



Gene Advocate

Issue 63

Rare Diseases

What is a rare disease?

Internationally, definitions vary among countries. Australia, for example, defines a rare disorder as a condition that affects about 2,000 people, while in the United States it is about 200,000 people.

In Europe, a disease or disorder is defined as rare when it affects less than 1 in 2000 citizens (Orphan Drug Regulation 141/2000). Rare diseases may affect 30 million European Union citizens.

Although by definition each rare disease occurs infrequently, collectively, approximately 8000 rare diseases affect approximately 6-10% of the population. This is equivalent to 30 million people in Europe, 25 million people in the US and 1.2 million Australians.

It is increasingly acknowledged that low prevalence does not equate with low impact. Although rare diseases vary considerably in aetiology, there are many commonalities. Rare diseases often have their onset in childhood, continue throughout life, are difficult to diagnose, are disabling and have significant impacts and burdens on the patient, family,

community and health services.

Despite this, rare diseases receive little attention. For many rare conditions epidemiological and scientific data are lacking, making the development of evidence based practice and policy impossible.

Many rare diseases are serious and/or chronic. Many are life-threatening. Even so, people with rare diseases often have trouble accessing the medical or other services they need because those making the decisions are not familiar with their diseases.

Characteristics of rare diseases

- Rare diseases are often chronic, progressive, degenerative, and often life-threatening
- Rare diseases are disabling: the quality of life of patients is often compromised by the lack or loss of autonomy
- High level of pain and suffering for the patient and his/ her family
- No existing effective cure
- There are between 6000 and 8000 rare diseases
- 50% of rare diseases affect children

- ✓ 80% of rare diseases have identified genetic origins. Other rare diseases are the result of infections (bacterial or viral), allergies and environmental causes, or are degenerative and proliferative.

Rare diseases are characterised by a broad diversity of disorders and symptoms that vary not only from disease to disease but also from patient to patient suffering from the same disease. Relatively common symptoms can hide underlying rare diseases, leading to misdiagnosis.

Rare disease patients face common problems:

- Difficulty in obtaining an accurate diagnosis
- Limited treatment options
- Little or no research being done on the disease
- Difficulty finding physicians or treatment centers with experience in treating a particular rare disease
- Treatments that are generally more expensive than those for common diseases
- Reimbursement issues related to private insurance, and Medicare
- Difficulty accessing medical, social, educational or financial services or assistance because those making the decisions are not familiar with the disease
- Feelings of isolation and of having been “forgotten” by our healthcare system.

How can things change?

- ✓ By implementing a comprehensive approach to rare diseases
- ✓ By developing appropriate public health policies
- ✓ By increasing international cooperation in scientific research
- ✓ By gaining and sharing scientific knowledge about all rare diseases, not only the most “frequent” ones
- ✓ By developing new diagnostic and therapeutic procedures
- ✓ By raising public awareness
- ✓ By facilitating the network of patient groups to share their experience and best practices
- ✓ By supporting the most isolated patients and their parents to create new patient communities or patient groups
- ✓ By providing comprehensive quality information to the rare disease community

Rare Disease Day February 28th 2011

This will be the fourth annual Rare Disease Day. The concept was launched in 2008 by EURORDIS, NORD’s counterpart in Europe. Last year, EURORDIS asked NORD to host Rare Disease Day in the U.S. In 2010, Rare Disease Day was observed in 46 nations around the world including Australia. The observance is always on the last day of February.

Information Source:

Australian Paediatric Surveillance Unit. <http://www.apsu.org.au/index.cfm?objectid=64B91B18-A313-7C8D-864397E5A22934D0>
Canadian PKU and Allied Disorders Inc. 4/6/2010. <http://canpku.org/pku-news/90-canada-not-fit-to-deal-with-rare-disorders>
Eurordis, Rare Diseases Europe. <http://www.eurordis.org/content/what-rare-disease>
NORD Rare Disease Day. <http://rarediseaseday.us>

Rare Disease Resources



The NORDD Rare Disease Support Community connects patients, families, friends and caregivers for support and inspiration.

This community is sponsored by the National Organization for Rare Disorders, an Inspire trusted partner.

Joining Inspire is completely free and usually takes less than a minute, visit: <http://www.inspire.com/groups/rare-disease/>

The Australasian Genetic Alliance (AGA)



The Australasian Genetic Alliance (AGA) facilitates networking

between groups, health professionals and patients and families to improve community and professionals knowledge of genetic conditions, their impact and available services.

The Alliance has taken a lead advocacy role in representing members interests in national genetics issues.

The AGA provides written and verbal submissions to government and others in the field of genetics and participates with like bodies nationally to further represent genetic support groups and their members.

AGA Member Groups

- Genetic Support Council WA (GSCWA)
- Genetic Support Network Victoria (GSNV)
- New Zealand Organisation for Rare Disorders (NZORD)

- ACT Self-Help Organisations United Together (SHOUT)
- The Association of Genetic Support of Australasia (AGSA)

Visit: www.australasiangeneticalliance.org.au

Genetic Support Groups



Our recently updated Directory of Genetic Support Groups & Community Support Organisations contains some information and details of rare genetic diseases and conditions.

The directory is available in PDF format on our website www.geneticsupportcouncil.org.au or you can ph 9485 8999 to order a hard copy.

Become a Rare Disease Day Partner or Ambassador

Rare Disease Day is about realizing the similarities among all individuals impacted by rare diseases. By working together we can raise awareness and create a global impact. Whether you are an individual or part of an organization, we want you to show your support by becoming a Partner or Ambassador.

Visit: <http://rarediseaseday.us>



Stem Cell Research: Scientists turn skin into blood

Stem cell researchers have found a way to turn a person's skin into blood, a process that could be used to treat cancer and other ailments, according to a Canadian study.

The method uses cells from a patch of a person's skin and transforms it into blood that is a genetic match, without using human embryonic stem cells, said the study in the journal *Nature*.

By avoiding the controversial and more complicated processes involved with using human embryonic stem cells to create blood, this approach simplifies the process, researchers said.

"What we believe we can do in the future is generate blood in a much more efficient manner," said study author Mick Bhatia of the McMaster's Stem Cell and Cancer Research Institute in the Michael G. DeGroote School of Medicine.

With the ability to create blood for transfusion from a person's own skin, the advance means someday patients needing blood for surgery or to treat anemia could bypass the blood bank and derive the necessary supply from themselves.

The breakthrough could also see future uses such as allowing patients undergoing chemotherapy to endure a longer regime of treatment without the breaks currently needed to rejuvenate the body.

Researchers have been able to perform the skin-to-blood

transformation in the past, but while using human pluripotent stem cells, widely known as embryonic stem cells.

Stem cells that are derived from human embryos hold significant promise for medical breakthroughs but also carry risks, such as the potential to create tumours.

But researchers say their new method can create enough blood for a transfusion from a four by three centimetre patch of adult human skin, and can avoid those potential hurdles.

Information Source & Full Article Available: SBS World News Australia. 8/11/2010. Available: <http://www.sbs.com.au/news/article/1405562/latest-from-wire/%20>

Additional Information

Scientists create blood from skin. Read the discussion and listen to the program from ABC Local Radio PM:

- <http://www.abc.net.au/pm/content/2010/s3060606.htm>

Direct conversion of human fibroblasts to multilineage blood progenitors.

Nature article available:

- <http://www.nature.com/nature/journal/vaop/ncurrent/full/nature09591.html>

Mandatory reporting of birth defects in Western Australia

After 30 years as a very successful voluntary Birth Defects Register, new regulations will commence in early 2011 that will establish it as a statutory register, to be known as the WA Register of Developmental Anomalies.

The decision to make this registry a statutory entity was made after extensive consultation with health professionals and health consumer representatives and provides legal protection for those making notifications. The information collected, the ways it is collected, the health professionals who notify cases and the manner in which the register operates are not expected to change.

For the purposes of the regulations, a developmental anomaly is defined as cerebral palsy or a structural or functional anomaly which is present at conception or occurs before the end of pregnancy and is diagnosed during pregnancy, after stillbirth, termination of pregnancy or after live birth but before 6 years of age. In the regulations, a developmental anomaly is prescribed as a condition of health, to which Part IXA of the Health Act 1911 applies.

The regulations include a section on the right of persons (or, in the case of children, their parents) to know if information about them is included on the register and, if so, to have a copy of that information. There is also a section allowing identifying information to be removed, once it

has been kept on the register for at least six years. Requests in relation to these two sections must be in writing. Non-identifying information may be retained.

More detailed information will be provided on the Office of Population Health Genomics (OPHG) and the King Edward Memorial Hospital websites early in 2011.

Source: Office of Population Health Genomics (OPHG) Newsletter
December 2010

Links

- OPHG Website:
www.genomics.health.wa.gov.au/home/
- King Edward Memorial Hospital Website:
www.kemh.health.wa.gov.au

Additional Information

About the Birth Defects Registry

- www.kemh.health.wa.gov.au/services/birth_defects/

Information about your Health Data
(2009)

- http://www.health.wa.gov.au/healthdata/docs/Health_data_brochure.pdf

Tribute to Australian Organ Donors

THE HON CATHERINE KING MP

Parliamentary Secretary for Health and Ageing

MEDIA RELEASE

18 January 2011

Tribute to Australian Organ Donors

A record number of 309 deceased Australians donated their organs and tissue for transplant during 2010, drawing praise and gratitude today from the Parliamentary Secretary for Health and Ageing, Catherine King.

The donations saved or significantly improved the lives of 931 Australians who were relying on a transplant. That transplant figure is the highest in more than a decade.

The figures were released today by the Australian and New Zealand Organ Donation Registry (ANZOD) and the Organ and Tissue Authority.

They show the 2010 deceased donation figures exceeded the total number of donations in any year in Australia's recorded donation and transplant history.

"This is a wonderful credit to those people who so generously donated and to their families who honoured the wishes of their deceased loved ones," Ms King said.

"It means improved health and life span for other Australians who were the beneficiaries of the donations and demonstrates that the message to encourage more people to discuss with their loved ones their wish to donate their organs and tissue is getting through.

"It also shows that the Government's \$151 million reform program to boost organ and tissue donation rates is on track and having an impact.

"As well as the community education program on organ and tissue donation, the reforms also include the establishment of a national clinical network of staff around the nation aimed at improving organ and tissue donation and raising awareness about organ donation in hospitals.

"I want to pay tribute to those Australians who have donated organs. They have left a living legacy. I also want to encourage all Australians to discuss with their families their donation wishes upon their death."

The figures released today show that the organ donation total of 309 equates to a national organ donor rate of 13.8 per million. This is significantly higher than the annual average of 205 donors that occurred during 2000 to 2008 (10.2 per million of population), prior to the implementation of the Australian Government's National Reform package in 2009.

"These figures are extremely gratifying and will bring renewed hope to those people on waiting lists for transplant surgery," Ms King said.

The full 2010 ANZOD figures are available at www.anzdata.org.au/anzod/v1/summary-org-donation.html

People wanting to know more about organ and tissue donation should visit www.donatelife.gov.au

Media contact – Peter Dwyer (Ms King's office) 0409 866 054.

Information Source: [http://www.health.gov.au/internet/ministers/publishing.nsf/Content/8B4749587E5E64EDCA25781C0003EC3D/\\$File/ck001.pdf](http://www.health.gov.au/internet/ministers/publishing.nsf/Content/8B4749587E5E64EDCA25781C0003EC3D/$File/ck001.pdf)

New test can screen couples for 448 diseases

A NEW genetic test could soon allow couples to screen themselves for hundreds of recessive diseases to prevent them passing them on to their offspring.



American researchers yesterday reported the new test was capable of detecting 448 inherited conditions such as cystic fibrosis, fragile X syndrome and sickle-cell anaemia.

The researchers, from the Drexel University College of Medicine, in Philadelphia, said couples could be screened before conceiving so they could make informed reproductive choices thereafter.

Advertisement: Story continues below For example, if a couple was told they had a 25 per cent chance of having a baby with cystic fibrosis, they could opt to use sperm donation or in vitro fertilisation to select unaffected embryos for implantation.

They could also have further screening during pregnancy at about 12 weeks with a view to abort an affected foetus. While such tests could significantly reduce the incidence of such diseases in the community, critics say they create unnecessary alarm about rare diseases and could lead to more abortions.

In a report on the finding in Science Translational Medicine yesterday, the researchers said their test would also be suitable to screen infants for treatable or preventable diseases where early diagnosis is beneficial.

The director of the Victorian Clinical Genetics Services, David Amor, said that although a similar genetic test to screen people for about 100 diseases became available on the internet last year, this test represented a new and more powerful tool

for accurately testing individuals for a very large number of recessive genes.

While many couples would be interested in using the test, the vast majority would not find any gene mutations to worry about, Associate Professor Amor said.

The test, which could be available to Australians within a year, raised many legal, ethical and practical issues, he said.

"While this testing is probably the way of the future, we haven't really got our heads around how to deal with it and how to adequately counsel patients about the testing," he said.

Information Source: Julie Medew. The Sydney Morning Herald. [Online]. 14/01/2011. Available: <http://www.smh.com.au/world/science/new-test-can-screen-couples-for-448-diseases-20110113-19pwo.html>

Additional Information

Gene test predicts 448 child diseases - Laiba Medical.

- www.laibamedical.com/pregnancy-and-child/568-gene-test-predicts-448-child-diseases.html

Gene test 'predicts 448 child diseases' - National Health Service (NHS) England.

- www.nhs.uk/news/2011/01January/Pages/dna-gentic-test-for-parents-before-pregnancy.aspx

The common problem of rare disease in general practice

Clinical Practice - The common problem of rare disease in general practice. Andrew W Knight and Timothy P Senior
MJA 2006; 185 (2): 82-83

Abstract

- ◆ Rare diseases affect 6%–10% of the population, which equates to about 1.2 million people in Australia having a rare disease.
- ◆ The United States, the European Union and many other nations have coordinated policies and patient advocacy groups for rare diseases as a group.
- ◆ Australia has enacted orphan drug legislation, but there is no coordinated approach either from government or from patient groups.
- ◆ General practitioners see rare diseases commonly, but their role for this group has not been adequately described.
- ◆ People with rare diseases and their families have similar experiences despite their different diagnoses. GPs are well placed to help with these problems.
- ◆ The development of a generic general practice strategy for these patients may improve their overall care.

In a combined total of 25 years of general practice experience, we have cared for patients with acromegaly, prolactinoma, Addison disease, glioblastoma, narcolepsy, corneal dystrophy, congenital hypothyroidism, listeriosis, juvenile-onset arthritis, Huntington disease, muscular dystrophy, syringomyelia, Marfan syndrome, Guillain–Barré syndrome, adamantinoma, Laurence–Moon syndrome, reflex sympathetic dystrophy, and the list goes on.

Why are we seeing so many rare diseases?

Rare disease is common. Estimates suggest that between 6% and 10% of the community suffer from a rare disease at any one time. This prevalence is similar to that of type 2 diabetes mellitus. There are significant funded strategies for rare diseases in Europe, the United States and other countries. What is happening in Australia, and what is the role of general practice in rare diseases?

Full Article Available: The Medical Journal of Australia (MJA). Available: http://www.mja.com.au/public/issues/185_02_170706/kni10328_fm.html#0_pgflid-1091794

Health of Australians with disability: health status and risk factors

Summary

Despite the overall improvement in population health, the gap between Australians with disability and those without disability remains large. In 2007–08, almost half (46%) of people aged 15–64 years with severe or profound disability reported poor or fair health, compared to 5% for those without disability.

People aged under 65 years with severe or profound disability had a higher prevalence rate of all types of selected long-term health conditions than people without disability. Almost half (48%) of people with severe or profound disability had mental health problems, compared to 6% of people without disability. The prevalence of physical long-term health conditions was higher for people with both mental health problems and severe or profound disability than for those with mental health problems but no disability.

People with severe or profound disability were more likely to have acquired a long-term health condition of the National Health Priority Areas (NHPAs) earlier than those without disability.

Among people aged under 65 years with severe or profound disability and mental health problems, the proportion who had behavioural and emotional problems with usual onset in childhood or adolescence was 14%, compared to 7% of those without disability.

Among people aged 15–64 years with a specific long-term health condition of NHPAs, the comparisons between people with severe or profound disability and those without disability showed that:

- the proportion who had diabetes or a high sugar level before the age of 25 years was 23% versus 7%
- the proportion who acquired arthritis before the age of 25 years was 14% versus 6%
- the proportion who first experienced osteoporosis before the age of 45 years was 43% versus 31%.

About 69% of adults aged 18–64 years with severe or profound disability were overweight or obese, compared with 58% for those without disability.

People aged 35–64 years with severe or profound disability who were overweight or obese were more likely than those without disability to report both cardiovascular diseases and diabetes (9% versus 1%).

Compared to people without disability aged 15–64 years, people of the same age with severe or profound disability were more likely to do a very low level of exercise or no exercise (43% versus 31%); to be current daily smokers (31% versus 18%); to start smoking before the age of 18 years (38% versus 22%); and were less likely to have never smoked (42% versus 61%).

About 42% of people aged 16–64 years with severe or profound disability had seriously thought about committing suicide, including 18% who had attempted suicide. This was associated with their high prevalence of mental and behavioural problems.

The Full Report: Australian Institute of Health & Welfare. Health of Australians with disability: health status and risk factors. Available: www.aihw.gov.au/publications/index.cfm/title/11608



Member Profile

The CHARGE Syndrome Association of Australasia is a not-for-profit organisation committed to improving the health and welfare of children and adults with CHARGE Syndrome. The Association supports families in Australia and New Zealand to enable children and adults with CHARGE Syndrome to reach their full potential.

The Association was formed in 1988 to assist families whose children were born with this rare condition. The Association subsequently became a registered company in 1996.

The condition was first identified in 1979 in the USA, the acronym CHARGE was given in 1981.

Since the formation of the parent support group in Australia, 1994, conferences have been held every 2 years. A newsletter is produced quarterly, fundraising activities occur and planning for future needs of these children and young adults is a major consideration.

Families of newly diagnosed children with CHARGE are provided with an information package on request, and parent and professional representatives are available to discuss CHARGE with any interested persons.

Interested medical and educational professionals have involved themselves in gaining data about children in Australia and New Zealand. Sych professionals also

keep abreast with current developments locally and internationally.

What is CHARGE Syndrome Definition

CHARGE Syndrome, also known as CHARGE Association, is a specific set of birth defects. The major diagnostic criteria were based on the letters in CHARGE. Not all individuals with CHARGE will have all the symptoms, and each of the symptoms can vary in their degree of severity. (Sandra L.H. Davenport, M.D.)

In 1979 Dr Bryan Hall first described the associated features of CHARGE. It became known as CHARGE Association when Dr Roberta Pagon coined the acronym in 1981.

Early Literature Acronym

In the early literature the acronym stood for:

- ◆ Coloboma of the eye
- ◆ Heart defect
- ◆ Atresia of the Choanae
- ◆ Retardation of growth and/or development
- ◆ Genital hypoplasia
- ◆ Ear malformations

The sub group of features included:

- ◆ Facial paralysis or palsy
- ◆ CHARGE facial features
- ◆ Central nervous system disorders

- ◆ Swallowing abnormalities
- ◆ Cleft lip and/or palate
- ◆ Urinary tract malformations
- ◆ Tracheo – oesophageal fistula

Recent diagnostic criteria

Dr Kim Blake et al in the Clinical Paediatrics Journal 1998, reclassified the major and minor characteristics of CHARGE Association.

Major Criterion:

- ◆ Coloboma
- ◆ Choanal atresia
- ◆ Characteristic ear abnormalities
- ◆ Cranial nerve dysfunction

Minor Criterion:

- ◆ Genital hypoplasia
- ◆ Developmental delay
- ◆ Cardiovascular malformations
- ◆ Growth deficiency
- ◆ Orofacial clefts
- ◆ Tracheo - oesophageal fistula
- ◆ Distinctive face

Despite these changes the acronym CHARGE remained.

More recently a team from The Netherlands, headed by Conny van Ravenswaay, claim to have identified the 'CHARGE Gene'. They detected in 10 out of the 17 individuals studied mutations in the gene CHD7.

Their paper is published in 'Nature Genetics Advance Online Publication' published online 8 August 2004.

What causes CHARGE syndrome?

CHARGE syndrome is a genetic condition, caused by a change (mutation) in a single gene, most often CHD7.

The gene is CHD7, located on the long arm of chromosome #8. It is a regulatory gene which plays a role in turning other genes on and off. Changes (mutations) in this gene have been found in more than half of all children with CHARGE tested to date.

In the vast majority, the mutation was new in the child - not detected in the parents. This confirms that CHARGE syndrome is a genetic condition caused by a new mutation in a dominant gene.

Further research is needed to find other genes that can cause CHARGE and to determine the function of the CHARGE genes in the developing fetus, babies, children and adults.

Diagnosing CHARGE Syndrome

CHARGE Syndrome (sometimes referred to as CHARGE Association) can only be diagnosed by a qualified Geneticist.

Further Information



CHARGE Syndrome Association of Australasia

Web: www.chargesyndrome.org.au

Information Source:

CHARGE Syndrome Association of Australasia. www.chargesyndrome.org.au/charge.html

US CHARGE Syndrome Foundation.
www.chargesyndrome.org/about-charge.asp



Coming Events

CONSUMERS REFORMING HEALTH 2011 CONFERENCE

Health Issues Centre will host the inaugural international conference for community engagement in health care.

Date: 18-20 July 2011

Public Forum on Sunday, 17 July 2011 at the Melbourne Convention and Exhibition Centre.

Consumers Reforming Health 2011 is a forum developed specifically for those with interest in involving consumers in health policy and process.

The conference upholds the essential principle that consumer participation is fundamental for health development, clinical governance, community capacity building and health outcomes.

The program aims to demonstrate and enhance this proposition by exploring the experiences, frameworks and practical strategies of participation in a number of health sectors among a diverse range of relevant stakeholders. It intends to broker some new learning on core principles for future change.

✓ Abstract submissions for the conference are now open.

Key dates:

- Abstract submission opens: August 2010
- Abstract submission closes: March 18th 2011
- Earlybird Registration deadline: April 1st 2011

<http://consumersreformhealth.asnevents.com.au>

Toyota Muscular Dystrophy Golf Classic 2011



Date: Friday 1 April 2011

Muscular Dystrophy WA (MDWA) is pleased to announce the 2011 Toyota MD Golf Classic. MDWA is very grateful for the ongoing major sponsorship received from Toyota WA and we look forward to another successful day.

The tournament will again be hosted by The Vines Resort & Country Club on Friday 1 April 2011.

Please make all enquiries to Mark Hullett at markmda@cyllene.uwa.edu.au or 0409 812476

Rare Disease Day 2011

Date: February 28, 2011

Rare Disease Day provides a wonderful opportunity to teach the public and professionals about specific diseases and to convey that rare diseases are an important public health concern receiving too little attention around the world.



<http://www.rarediseaseday.org>

International rare disease symposium

Awakening Australia to Rare Diseases: Global perspectives on establishing a coordinated approach to a national plan.

Date: April 18th to 20th 2011

Currently, Office of Population Health Genomics OPHG is co-planning a rare disease symposium from April 18th to 20th, 2011 in Fremantle WA.

This symposium will provide:

- Information to develop a framework for a national plan
- Opportunities for community and professional engagement
- Opportunities to build long term relationships.

The symposium will attract health, disability and social service providers, patient support organisations, patient advocates, researchers, biotechnology and pharmaceutical industry representatives and government policy makers.

More information will be available from the symposium website www.raredisease.com.au

National Epilepsy Awareness Month. Epilepsy Australia

Date: March 26 2011

March is National Epilepsy Awareness Month, which includes a range of events, community activities and media campaigns around Australia to raise awareness and an improved understanding of epilepsy. Events will culminate in Purple Day for Epilepsy Awareness

when everyone is encouraged to wear purple for epilepsy.

Epilepsy Association of Western Australia

Ph (Office): 08 9346 7699

Helpline: 1300 37 45 37

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

National Website: www.epilepsyaustralia.net

World Down Syndrome Day



Date: March 31st 2011

People with Down syndrome love to celebrate, so it is apt that a day be set aside for people with Down syndrome to celebrate their lives and for people all over the world to celebrate with them.

World Down Syndrome Day is a great opportunity to raise awareness about people with Down syndrome and to highlight individuals with Down syndrome: their strengths, their achievements, what they have to say, their active involvement in the community, their personal contributions to the community and just who they are!

It is also a great opportunity to acknowledge all the people in the community who support people with Down syndrome and who share their lives in some way.

For further information visit 'News & Events' at: <http://www.dsawa.asn.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

How Australians view their health care system: Australian Health Survey 2010 Results

To find out how Australians view their health care system, the Menzies Centre for Health Policy and The Nous Group (Nous) conducted a telephone survey of a representative sample of 1201 Australians in July 2010.

The survey asked questions on the following subjects: satisfaction with the health system; support for health reform; access to health care services; confidence in services and affordability of health care. The preliminary report is provided via the link below.

See: www.menzieshealthpolicy.edu.au/mn_survey/index.htm

Trust It or Trash It?

This is a tool to help you think critically about the quality of health information (including websites, handouts, booklets, etc.). Click on Who said it? When did they say it? and How did they know? to guide you through the process.



Visit: <http://www.trustortrash.org>

Does it run in the family?

A FAMILY HEALTH HISTORY TOOL FROM GENETIC ALLIANCE

This tool helps you create personalized booklets to start conversations about health in your family and community.

The sooner we understand the link between our family's health and our own, the sooner we can make changes and take steps to live longer, healthier lives. A peek into the past can reveal a lot about your future.

Family health history:

- Leads to conversations about health
- Helps healthcare providers diagnose conditions
- Tells you about your risk for specific diseases

Visit: <http://www.familyhealthhistory.org>



Better Boards - Assessing and Improving your Board

Help Sheets Part 3

Even well-functioning Boards will need some fine-tuning from time to time. It is essential, therefore, that all not-for profit Boards take regular steps to assess how they are performing and work to improve themselves.

- How effective is my Board?
- Why do we need to assess our own performance?
- How can we tackle this task sensitively?

These help sheets will help you answer these questions and ensure your Board continues to think about and improve its performance without breaking stride.

We will be adding new help sheets regularly so keep coming back to check out the latest tools and resources to help strengthen your Board and yourself as a Board member.

Visit: http://www.ourcommunity.com.au/boards/boards_article.jsp?articleId=1301

Health Consumers' Council Your Voice on Health

MAKING A COMPLAINT

Information for Health Consumers

The online leaflet gives options for what you can do if you are concerned about a health service.

Visit: [http://www.hconc.org.au/publications/02%20Leaflets/HC27395_DL_Complaint_hires\[2\].pdf](http://www.hconc.org.au/publications/02%20Leaflets/HC27395_DL_Complaint_hires[2].pdf)

Micro-array testing brochure Centre for Genetics Education

A new brochure on a special genetic test known as micro-array testing. This test is part of an investigation into the developmental or health concern you have about your child or yourself and will look for extra or missing DNA segments.

Download PDF: <http://www.genetics.edu.au/pdf/microarraybrochure.pdf>

Affordable Computers GreenPC

GreenPC is a non-profit social enterprise that is part of Infoxchange Australia's vision to help bridge the digital divide and ensure all people have equal access to information technology.

GreenPC refurbish computers donated by corporate and government organisations. Each computer is thoroughly cleaned, tested and updated before being offered for sale to community groups, low income individuals (must be holders of a current Health Care Card, Aged Pension Card, Disability Card or some other form of documentation relating to low income or disadvantage) and the general public. GreenPC also offers computer systems and information technology services at affordable rates to registered charities and community groups.

Visit: <http://www.greenpc.com.au>
GreenPC Western Australia
Ph: 9434 0530



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Grants

Google Grants

Closes: Ongoing

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.

What is 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.

Aim: To award non-profits with free AdWords advertising on Google.com.au to raise awareness and increase traffic.

- ✓ Each organisation choosing to participate in Google Grants receives at least three months of free advertising.

Limitations:

- ✓ You will need to have a website to advertise with Google Grants.

- ✓ You must be a charitable organisation with Deductible Gift Recipient (DGR) Status to apply for this grant

Further Information

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

Program guidelines and details: <http://www.google.com.au/grants/details.html>

Email: googlegrants-australia@google.com

ING Foundation Grants

Closes: two funding rounds - 1 March and 1 September 2011

The ING Foundation makes financial grants to organisations that have a strong focus on supporting children in acute need (and/or their immediate families).

- There are two funding rounds, closing on 1 March and 1 September, each year.
- The Board of Trustees for the ING Foundation meet twice annually to consider applications for financial assistance.
- Grants range from a few hundred dollars up to \$15,000.00 per project.
- Applicants may apply for a grant in both funding rounds in one year. If your

application is successful however, you may not apply for funding from the ING Foundation again for 12 months. For example, if you received a grant for financial assistance in September 2009 you are not eligible for funding until September 2010.

Selection Criteria

Applications must meet all of the selection criteria below to be considered for an ING Foundation grant:

1. Must be registered with the Australian Taxation Office as an approved charity;
2. Provides services or resources that directly benefit individuals in need;
3. Deals with welfare, disabled or the disadvantaged; and
4. Assists children (and/or their immediate families)

Grants will not be awarded for:

- International grant applications
- General administration or infrastructure
- Communication tools (eg brochures) or office equipment
- Bricks and mortar projects
- Sponsorship of individuals
- Assistance with fundraising projects
- Support for foundations that allocate funds to other charitable organisations

Further Information

Website: <http://www.ingfoundation.com.au>

Online Application Form: http://www.ingfoundation.com.au/main.php?page=grant_application#

Email: info@ingfoundation.com.au

Community Aids and Equipment Program (CAEP) Disability Services Commission (WA)

Closes: Ongoing

Aim: The Community Aids and Equipment Program (CAEP) aims to provide an equitable accessible and consistent statewide scheme for the provision of equipment and home modifications for the benefit of people with a long-term disability, who are eligible for services from the Commission and living in the community.

All equipment and home modifications funded by CAEP are provided not only to enhance the function, independence and safety of the equipment user, but also to assist carers in their caring role.

See your GP to access CAEP

Ask your local GP to check your eligibility and refer you to your local CAEP service provider. Online resources are available to assist your GP.

Further Information

Contact Your Local Area Coordinator.

TTY: 9426 9315

Freecall: 1800 998 214

Website: <http://www.disability.wa.gov.au/forindividuals/disabilityservices/aidsequipment.html>

Email: caep@dsc.wa.gov.au



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'.

Opitz G/BBB syndrome

A Western Australian family living with Opitz G/BBB syndrome are seeking contacts with others living with this condition.

Synonyms of Opitz G/BBB syndrome:

- BBBG Syndrome
- Hypertelorism with Esophageal Abnormalities and Hypospadias
- Hypertelorism-Hypospadias Syndrome
- Hypospadias-Dysphagia Syndrome
- Opitz BBB Syndrome
- Opitz BBB/G Compound Syndrome
- Opitz BBBG Syndrome
- Opitz G Syndrome
- Opitz Hypertelorism-Hypospadias Syndrome
- Opitz Oculogenitolaryngeal Syndrome
- Opitz-Frias Syndrome
- Telecanthus-Hypospadias Syndrome

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

Expressions of Interest: Dyspraxia WA Support Group

The Australian Dyspraxia Association are wishing to commence a new support group in WA but unfortunately do not have a person to train as the volunteer mentor. If you are a member of the ADA and have a few hours spare each month, have a child with Dyspraxia and would like to meet with and support others in the same boat, please contact Christine to discuss this matter further. Training will be provided. If you would like to join this support group, please contact Christine at Head Office.

For further information, please email: information@dyspraxia.com.au



GSC Members

Full Members

Acoustic Neuroma Association of Australia, WA Branch
Alzheimer's Association of WA
Angelman Syndrome Association
Arthrogryposis Support Group
Australian Cystinosis Support group
Australian Dyspraxia Association
Australian Huntington Disease Association
Australian Leukodystrophy Support Group
Australian Mitochondrial Disorder Foundation
Australian Pituitary Foundation WA Branch
Australian Tuberous Sclerosis Society Inc.
Australasian CHARGE Syndrome Association
Charcot-Marie-Tooth Association Australia
Cleft Lip and Palate Society of WA
Coeliac Society of WA
Cornelia De Lange Syndrome Support Group
Cushing's Disease Support Group
Cystic Fibrosis WA
Diabetes Australia - Western Australia
Down Syndrome Association of WA
Dyslexia SPELD Foundation WA Inc
Epilepsy Association of WA
Even-Keel Bi-Polar Support Association (Inc)
Familial hypercholesterolaemia Support Group WA
Fragile X Support Group WA (Inc)
Haemophilia Foundation WA Inc.
Heart Kids WA
Klinefelters Support Group
Learning and Attention Disorders Society of WA (LADS)
LQTS Support Group WA
Lupus Group of WA
Lymphoedema Association of WA
Mental Illness Fellowship WA
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
Perth Tourette Syndrome Support Group
PXE Support Group of 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
Turner Syndrome Association of Australia (WA Branch)
Support Organisation for Trisomy and Related Disorders of WA (SOFTWA)
Usher Syndrome Support Group
Western Australian Retinitis Pigmentosa Foundation

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
Myasthenia Gravis WA Friends and Support Group
Office of Population Health Genomics
People with Disabilities WA
The Chromosome18 Registry & Research Society
The Kalparrin Centre
The Neurological Council of WA Inc
Western Australian Deaf Society Inc.

Individual Associate Members

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

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



Our Location and Mailing Address:

Suite 7, Oasis Lotteries House
37 Hampden Rd, Nedlands WA 6009

Attach mailing
address label here

Contact Us

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ABN: 63 614 315 270

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37 Hampden Rd, Nedlands WA 6009

Phone: 08 9485 8999

Email: info@geneticsupportcouncil.org.au

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Kristina Sengotta - Resource Officer
Joan Ryder - Book Keeper



GSCWA

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