



Australasian Genetic Alliance (AGA) Newsletter March 2009

Welcome

Welcome to the first Australasian Genetic Alliance (AGA) Newsletter. As this is our first newsletter, we have taken the opportunity to introduce ourselves.

We hope you enjoy this newsletter and welcome your feedback and ideas for further updates.

Kind regards, Leah Lonsdale,
*AGA Chair and
Coordinator, GSNV
(Honorary Secretariat)*

Who is the AGA?

There are more than 1000 Genetic Support groups in Australasia able to provide support and information to individuals and families living with a genetic condition. A support group can offer the opportunity to reduce isolation, share experiences, practical suggestions and ways of coping. The Australasian Genetic Alliance (AGA) facilitates networking between these groups,

health professionals and patients and families to improve community and professional 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.

How do I have my say?

Please feel free to contact us with your ideas, issues and suggestions. We also appreciate you bringing issues to our attention. You can contact us through your local member organisation (which can be located at <http://australasiangeneticalliance.org.au> and click "database") or via the Secretariat as listed below. The Secretariat contact person (and Chair) is Leah Lonsdale at the Genetic Support Network of Victoria till May 2009.



Supporting people living with a genetic condition

Increasing community awareness by networking & sharing resources

Representing common interests

Australasian Genetic Alliance

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Website: www.australasiangeneticalliance.org.au Email: info@australasiangeneticalliance.org.au

Member Organisations:

- Self-Help Organisations United Together (SHOUT) – Australian Capital Territory
- Genetic Support Network Victoria (GSNV) – Victoria
- The Association of Genetic Support of Australasia (AGSA) – New South Wales
- Genetic Support Council WA (GSCWA) – Western Australia
- Self Help Queensland (SHQ) – Queensland
- New Zealand Organisation for Rare Disorders (NZORD) – New Zealand



Rare Disease Day
28 February 2009

www.rarediseaseday.org

The 28th of February is Global Rare Diseases Awareness Day. ***Did you know that rare diseases are actually common?*** There are about 8000 rare diseases affecting an estimated 1.5 million Australians, including about 300,000 Australian children.

The Australian Paediatric Surveillance Unit convened a rare diseases meeting in Sydney on the 9th of February 2009 to discuss the need for a more co-ordinated approach to providing health care and support for Australians affected by rare diseases. The meeting was attended by a group of eminent researchers, health professionals and family support groups, including representatives from the European Rare Diseases Organisation www.eurordis.org and the New Zealand Rare Diseases Organisation www.nzord.org.nz. We discussed the need for an integrated patient support organisation for Australians affected by rare diseases, improved access to information and education for health professionals, and new models of healthcare.

Discussions highlighted the striking similarities among the ~8000 rare diseases:

- Most begin in childhood (usually diagnosed in children aged < 2years) and continue throughout life
- Obtaining a definitive diagnosis is often difficult and delayed
- Many rare diseases have no cure or require new high-tech and often expensive treatments
- Neurological and intellectual disabilities occur in about half of all cases regardless of disease type and lead to reduced independence and opportunities
- Families experience isolation, psychological and financial stress

- Health professionals have inadequate access to information, education and resources
- It is estimated that about 35% of deaths in children aged < 1year are due to rare diseases.

The European Union, Canada, the UK and New Zealand have recognised the complex problem of rare diseases and are responding by establishing national support organisations for rare diseases, by making rare diseases a public health and a research priority and by developing integrated clinics that pool expertise to enable quicker diagnosis and better access to treatments and interventions. A Creswick Foundation Fellowship will allow Dr Zurynski to study health and community services for rare diseases operating overseas and to implement this knowledge in Australia.

The second Rare Diseases Global awareness day was held on the 28th of February 2009 and Australian organisations such as the Smile Foundation www.smile.org.au and Association of Genetic Support of Australasia (AGSA) (www.agsa-geneticsupport.org.au) held events to highlight the importance of rare diseases. The Australian Paediatric Surveillance Unit www.apsu.org.au has joined the global effort to raise awareness of rare diseases: www.rarediseases.org

The AGA supports the principles in the International Genetic Alliance's position statement on Rare Diseases (<http://www.internationalgeneticalliance.org/english/View.asp?x=1441>)



UPDATES FROM AROUND AUSTRALASIA

Organisations listed by country, in alphabetical order. We look forward to the development of other Australasian peak bodies throughout Australasia and invite them to join the Alliance, to help make us truly representative of the region.

AUSTRALIA

GENETIC SUPPORT COUNCIL WA (GSCWA)

The GSCWA has three main areas of focus:

- Act as the peak body in WA through which member organisations participate and cooperate with relevant state, national and international bodies to raise awareness of issues for those living with or affected by genetic disease and to promote advocacy, leadership and cooperation between member groups and community organisations within the human genetic services field;
- Provide support and advocacy to newly diagnosed individuals and families to ensure they have access to up-to-date information about resources and support services that may be of benefit including referral to genetic support groups; and
- Facilitate the development of genetic support groups by providing expertise, information and technology including access to office facilities and assist with the development of resources such as information packages, pamphlets and design and printing of promotional materials.

Our current activities include consultations with members to explore some of the complex and challenging issues that emerge from the governance of biobanks, direct to consumer (DTC) genetic testing and the Australian Medical Council (AMC) draft code of professional conduct for doctors, 'Good Medical Practice'. We are also working with families, individuals and health professionals to establish a Familial Hypercholesterolaemia (FH) support Group in WA. 1 in 500 West Australians have FH but 80% of cases are not diagnosed. The aim of establishing a FH support group is to improve information, communication and support services for families with FH and help prevent premature deaths caused by high cholesterol and cardiovascular disease.

Further information about our work can be found on the GSCWA website at www.geneticsupportcouncil.org.au or by contact our office on (08) 9389 6722. We welcome your enquiries.

Genetic Support Network Victoria (GSNV)

GSNV is a community resource for people affected by genetic conditions and their families, condition-focused support groups, health professionals, government and the general community.

GSNV empowers people to overcome genetic challenges by:

- providing a friendly, informed point of contact
- linking people and groups in with appropriate services

- Connecting people facing similar challenges ('in the same boat')
- Hosting events such as family days, workshops and seminars
- Representing people with genetic conditions at a State and National level by sitting on key committees and making submissions

Recent GSNV initiatives include:

- *Legal Issues for Carers* workshop on Sunday 22nd March
- Youth Action Challenge (19th April)



- Genetic Support Awareness Week (19th – 25th April)
- Developing a Face Book cause to be more accessible to young people
- Facilitating two seminars as part of the Masters for Genetic Counselling course by Melbourne University and
- Submissions including:
 1. NHMRC's review of its guidelines on disclosing genetic information to relatives,
 2. Input into the evaluation of the DHS Birth Defects Register subcommittee, and
 3. DHS Strategic Framework for Paediatric Services in Victoria review.

GSNV has been delighted to act as AGA Honorary Secretariat for the past two years.

To find out more about GSNV please visit our web site (www.gsnv.org.au) or contact us by phone on (03) 8341 6315.

Self Help Queensland (SHQ)

SHQ is undertaking two major projects in 2009. One is a free, online searchable Directory of Queensland Self Help and Support Groups which will replace the now outdated hard copy version. The Directory will be available free to everyone to search for groups, and will form part of a brand new website containing a broad range of user friendly functions, fact sheets for self help groups and professionals, chat etc.

The second project, called 'Mercury Rising' is an exciting endeavour funded with a grant of \$300,000 from Disability Services Queensland to help build the capacity of the mental health self-help sector across the State. It is our hope that the success of this project will be a strong force in helping us convince Queensland Health to extend a similar opportunity to other groups, particularly genetic support groups.

SHQ continues to provide information and referrals to genetic support groups, and produces a quarterly newsletter which features a very popular column by Dr Kim Summers on topical genetic issues, as well as providing opportunities for genetic groups to promote their group and educate the broader community.

SHQ is a founding member of AGA, and is currently participating in AGA's important bid to develop a national approach to rare diseases. Because SHQ is a generic clearinghouse, it differs from the other AGA members in that its focus is on the broader self help sector. While SHQ is concerned with the sustainability of genetic support groups, it does not have a mandate from groups or individuals with genetic conditions to comment or vote on specific policy issues pertaining to genetics. SHQ contributes to AGA as best it can within these limitations.'

Association of Genetic Support of Australasia (AGSA)

AGSA is a tax-deductible registered charity providing support and information for individuals and families affected by a genetic condition so rare they do not have their own support group. AGSA has a Contact Register representing over 800 genetic conditions and is funded by NSW Health. AGSA aims:-

- Provides a contact point for families who are affected by genetic conditions so rare that they do not have their own support group.
- Facilitates access to individual support groups for those families with particular genetic disorders.
- Provide a forum for the exchange of information between support groups regarding available community services
- Educate the medical and allied health professionals and the community about genetic disorders.
- Consult with government bodies both Federal and State for appropriate funding for genetic services.



AGSA has a number of projects.

- Facilitating contact with others with the same or similar condition
 - AGSA runs 3 genetic seminars a year to assist families with information and to provide informal contact with each other often establishing a support group with peers willing to be the contact person for new families.
 - Rare Treasures Support Group representing 250 chromosomal abnormalities working in partnership with Unique in the UK.
 - Filling the Void Project for Carers supporting people living in isolation who are caring for someone with a genetic condition. This involves telegroup counselling, outreach rural seminars and sibling workshops and individual counselling with a psychologist.
 - BRCA1/2 and unknown gene faults information day has been running for 8 years sponsored by the NSW Cancer Institute.
- AGSA is the consumer representative on the NSW Newborn Screening Committee and the NSW Genetics Advisory Committee and the Acting President of the International Genetic Alliance.
 - Lobbying to have Congenital Adrenal Hyperplasia on newborn screening and raising awareness of rare diseases through Genetic Awareness Week and Rare Disease Day working with the Australasian Paediatric Surveillance Unit as part of their taskforce. (See page 2).
 - NZ-Australia Genetics Policy Roundtable 2008 – ‘Transforming Research into Practice, Policy and Law Reform’.

A calendar of events for 2009 is available from the AGSA office. Please call Dianne on +61 2 9211 1462 for your copy.

NEW ZEALAND

New Zealand Organisation for Rare Disorders (NZORD)

In New Zealand there has been a lot of work in the past few years on the development two major strategies – a Carers strategy and a medicine strategy. These have both been completed and adopted by government. NZORD played a leading role in the support group networks that joined together to push for them. They are high level documents that indicate future directions and objectives for government, so more work will be needed to ensure there is effective implementation of them.

Another major piece of work for 2009 and beyond, is working with officials in the Ministry of Health and District Health Boards to improve diagnosis and clinical care for patients with rare diseases. Most countries

have a tradition of relative neglect of rare genetic conditions and New Zealand is certainly no exception to this. Our action plans includes lobbying for an upgrade to our genetic services, and the development of specialist clinics to oversee the health care of those with the most complex disorders.

NZORD has learnt a lot about these issues through its contacts with AGA and other international networks, and we will continue working together to increase our knowledge and improving health outcomes and support for families affected by rare genetic diseases. *Check out our activities at www.nzord.org.nz*