

AGSA

THE ASSOCIATION OF GENETIC SUPPORT OF AUSTRALASIA INC.

NEWSLETTER

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MISSION STATEMENT

To facilitate support for
Those affected directly or
indirectly by genetic
conditions throughout
Australasia.

Dear All,
Here is a date for your diary -
AGSA's AGM will be held on
Saturday 27th October 2007 at
11.30 am at AGSA.

Recently AGSA organized a
national Williams Syndrome
Conference in conjunction
with Macquarie Centre for
Cognitive Science, Macquarie
University and the Williams
Syndrome (IHC) Association
Inc. I am pleased to say it was
a huge success. Over 120
people attended and 72
families participated in
research by giving blood,
volunteering for 3D imaging
and eye movement studies.
Prof. Peter Hammond, a
Professor of Computational
Biology in the Molecular
Medicine Unit at UCL's
Institute of Child Health in
London and **Dr May
Tassabehji** - Academic Unit
of Medical Genetics,
University of Manchester, St
Mary's Hospital, Manchester,
UK were the keynote
speakers.

A Williams syndrome
brochure was produced for
the conference and it will be
distributed to all clinical
genetic departments in
Australia. This would not have
been possible without the
generous collaboration of the
New Zealand WS support
group who kindly modified

their WS brochure for
Australia.

AGSA held the annual Breast
Cancer Information day on
14th October 2007. This was
kindly sponsored by the
Cancer Institute of NSW. This
event was well attended with
over 70 attendees from all
over NSW. A group from
Newcastle got together and
travelled down in a friend's
bus.

Genetic Awareness Week
launch was good fun where
Peter Allison, author of
"Whatever you do don't run",
entertained us with his stories
of Africa. Prof Ron Trent, and
Lynne Foxall gave stimulating
talks and we were given a
glimpse into genetics of the
future. I would like to thank
Prof David Sillence for his
excellent job of chairing the
evening.

Sadly Laurie Taylor will be
leaving AGSA on 24th October
2007. I would like to wish
Laurie every success for the
future and to thank her for her
tremendous contribution to
Filling the Void Project.

Until next time
Dianne Petrie

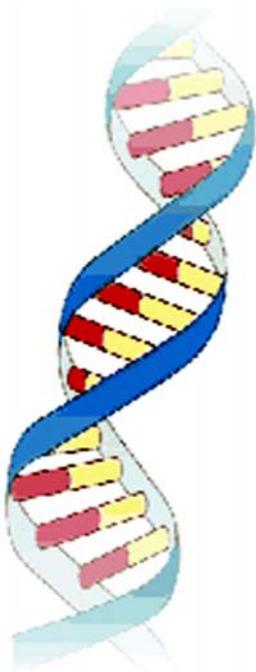
FILLING THE VOID REPORT

Hi all, I hope that the year has been treating you well and that you are enjoying the school holidays.

It's been an incredibly busy year with the FTV project – I thought I would give you a quick overview.

- Seminars & Sibling workshops in Ballina and Orange
- Telegroup counselling (TGC) – 1 dad's group, 1 mum's group and 1 sibling group
- 2 weekends away for parents and unaffected siblings at Yarrabin
- upcoming seminar and sibling workshop for individuals / families affected by Double Y syndrome and Klinefelter syndrome.

Unfortunately, I will be finishing at AGSA at the end of October. It has been a privilege and an honour to work on this project and I would like to say a big thank you and farewell to all of the families I have had the pleasure of meeting and getting to know. Best of love and luck to you all. **Laurie Taylor**



CONFERENCES

Gold Conference

Hunter Genetics, Newcastle NSW
Friday 2nd November, 2007
11.00am to 5.00pm

Meeting will be held at the Crowne Plaza Hotel, on the waterfront of Newcastle Harbour

Workshop on the newly identified genes causing XLMR (X-linked mental Retardation) following the October 2007 12th International Meeting on fragile X and XLMR in Venice.

The programme includes invited speakers Prof Jozef Gecz, who will give an overview of XLMR, and Prof Ingrid Scheffer, who will talk on finding the gene and clinical features of EFMR (the newest X-linked cause of MR and epilepsy in females). Dr Anna Hackett, will be talking on the availability of testing.

The meeting will be "back-to-back" with the Advances in Tuberosclerosis meeting in Sydney on 3rd November. Transport by car, at no cost, can be arranged from Newcastle to Sydney for that meeting.

A dinner will be arranged for Friday night. Registration (lunch included) \$50.00 – due date 26th October

Post to GOLD Service
P O Box 84
WARATAH NSW 2298
Email: margaret.jobber@hnehealth.nsw.gov.au

“Advances in Tuberosclerosis- From Pathway to Therapy”

The Australasian Tuberosclerosis Society in conjunction with Sydney Children's Hospital Randwick is holding a Medical/Family Conference.

When: Saturday 3 & Sunday 4 November 2007
Where: Sydney Children's Hospital, Randwick
Enquiries & Registration:

Families
Ph: 1300 733 435
Email: info@atss.org.au
Website: www.atss.org.au

Medical Professionals
Ph: 02 9382 1708
Dr David Mowat d.mowat@unsw.edu.au
Dr John Lawson
John.Lawson@unsw.edu.au



Fabry's Support Group

Inaugural Conference. Please join us in Brisbane for your Fabry's Support Group Conference **27th & 28th October 2007**

Venue: Oaks North Quay Hotel, Brisbane

You will remember last year we surveyed our members to find out what you want... the results were overwhelming... So here it is, your first conference dedicated to Fabry's.

We are hoping as many of our members as possible will join us to make this a rewarding experience for all. We have secured the services of some of Australia's leading experts in Fabry's disease to speak to our group. We hope you will take this opportunity to join your fellow members for a great weekend. We have lots happening to make your experience with your support group both informative and enjoyable

Here's a sneak peak of what we have planned:

Saturday: 27th October, 10:00am - 12:00noon

Registration/Meet and Greet

1.00pm Conference Commences

Professor David Sillence - Department of Medical Genetics, Westmead Hospital

Update on Treatments for Fabrys

Dr Phillip Juffs - Medical Team Leader Nutrition and Dietetics Royal Brisbane & Womens Hospital

Nutrition and Fabry's

7.00 pm Dinner—sponsored by Genzyme, Guest Speaker Lyn Fidler

Sunday: 28th October

10:00am Professor Jack Goldblatt - Director Genetic Services of WA

Women and Children with Fabry's Disease

11:00 am Fabry's Support Group AGM

12:00 Conference Concludes

EARLYBIRD OFFER - We have booked a limited number of rooms for FSG members for the Saturday night, get in early and your accommodation could be FREE. We need your commitment to attend by the 10th August 2007 and your room will be paid for, all you need to do is get yourself to Brisbane. Please Note: you must be a financial member of Fabry's Support Group to be included in this offer. Limited to the first 20 rooms.

HGSA Annual Scientific Meeting 2008

The 2008 Human Genetics Society Annual Scientific Meeting will be held 2-7 August 2008 at the Stamford Grand Hotel in Glenelg - a popular beachside suburb of Adelaide.

CONTACT CORNER

AGSA will publish requests for contact and letters from people searching for families with similar experiences, from those seeking or contributing specific information as well as other resource information.

Anyone who wishes to reply to a request or a letter should write direct to the individual or group concerned where an address is provided. The AGSA office may be contacted for the information to be passed on in the case of anonymous requests. Privacy and anonymity will be ensured if requested.

While AGSA aims to facilitate contacts between families it is unable to assess the suitability of these in individual cases.

It should be remembered that a shared genetic condition does not mean an equally shared value system between families. Different degrees of acceptance and different mechanisms for coping will be encountered and a non-judgmental approach is recommended in establishing contact.

Prune Belly syndrome

"I am a 38 year old female and have this condition. I know of only one other person in Australia who lives in Qld. I am therefore searching for other people with the condition." Please contact AGSA for details.

Maternal uniparental disomy of chromosome 14

"Our 11 month old granddaughter has been diagnosed with MUPD14 and I would like to know if there are any members who have registered who may be able to share information as I am led to believe this is a very rare chromosome diagnosis." Please contact AGSA for details.

How to cook a tasty meal with limited ingredients?

Attention: Frustrated Parents.

I have two little boys with many allergies and sensitivities to a lot of commonly used food, like milk, egg, nuts, tomato, and potato and fish, to name just a few. Over the past year, I've written down many of my wild creations and now at last my booklet is finished.

My little compilation of 14 easy recipes will start you off on a wonderful new discovery of basic food and flavour using just 35 basic ingredients.

If you are sensitive or allergic to almost every food under the sun, give my recipes a try. Bulk orders for just \$1 per copy



Individual booklets \$2 per copy
Postage \$1 anywhere in Australia, overseas to be advised.

Please allow 30 days for delivery.

Contact: gretakel@hotmail.com.au

for inquiries and orders

Workshops/service providers

Miroma - 9337 5167

Windgap - 9693 2399

Disability Services Australia - 93102055

Ability Options - 98346244

The Spastic Centre of NSW - 9451 9022

Job Support - 9554 9555

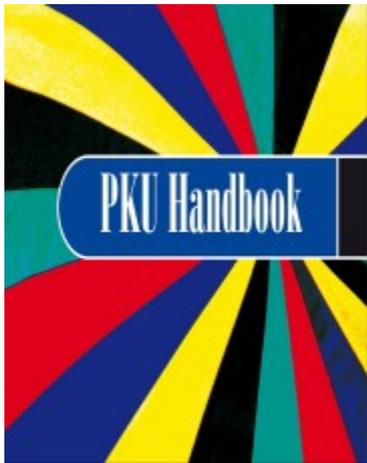
AFFORD - 9727 9199

Civic Training Services - 9524 8755

Jewish Care - 9331 5184

Society of St Vincent De Paul - 9519 3044

PKU Handbook



The PKU Handbook was prepared by the Dietitians Working Party of ASIEM in 2005 and can be downloaded as a .pdf file from the Human Genetics Society of Australasia website. <http://www.hgsa.com.au>

GENETIC AWARENESS WEEK PRESENTATION

Challenges Ahead in Human Genetics

Slides only of presentation

By Ronald J.A. Trent

Dept of Molecular & Clinical Genetics,
RPA Hospital, Sydney South West Genetics Service, & Central Clinical School, University of Sydney

Where are we now with human genetics?

Human Genome Project – completed. This sets the template for genetic discoveries, genetic knowledge and applications for the future.

We are now in the **Genomics era** - no longer do we deal only with single gene effects e.g. cystic fibrosis, but the technology now allows multiple genes (i.e. hundreds to thousands) to be studied.

Genomics opens up the option for assessing **gene environment interactions** – prevention becomes possible.

Focus on the traditional Mendelian single gene disorder e.g. haemophilia, cystic fibrosis has moved to more complex diseases = many genes +environmental interactions.

Distracting for some of “the less common single gene disorders that are of particular interest to us”

Challenge - Technology

To a large extent technology is now the driver for development of – hypothesis driven research. Therefore, we can expect newer, better and easier way to do genetic tasks e.g. sequencing DNA and the \$1,000 genome.

How is this technology regulated or controlled – because a lot of it is coming from industry?

Technologic advances – driven by industry
“Genetics and biomedical technology open up vast avenues for research and can provide humankind with much needed therapeutic tools.

But, where human life and dignity are at stake, technology cannot be left to govern ethics....” WHO 1995



The traditional laboratory now directly approaches the patient cutting out the doctors.

Challenge - The Internet.

Can the internet be regulated?

DIY or direct-to-consumer DNA testing. Now readily available to a wide range of "clients" e.g. nutrigenetics, dermatogenetics, "life-style" choices e.g. Personalised Diet Plan You Own DNA is just a step away.

e.g. Holland's Cells4Health Clinic

www.cells4health.com

Implications of the Internet for modern genetics



eCounselling
eConsulting
eHealth records
Communication
Education
Choices
Accessibility

Regulation
Standards
Education
Support
Liability
Access

Challenge - Workforce and service delivery

1. Genetics traditional model when it comes to service delivery – clinics, face-to-face consultations, laboratory testing etc. Unlike traditional disciplines the knowledge base in genetics changes considerably, and the majority of laboratory testing is not funded through Medicare.

2. Workforce issues are similar to other disciplines – not enough medical graduates and not enough genetic counsellors etc.

Genetics does not close the ED, does not require beds or lead to long waiting lists. Difficult to raise profile in terms of competing priorities

Challenge: Genetic type Therapies

Traditional drugs now supplemented by designer drugs (based on DNA basis for disease)

New type of therapeutics now coming onto the market – "cellular therapies" – includes gene therapy, stem cell therapies & perhaps xenotransplantation

Add nanotechnology – and the future is unlimited.

Challenge - Role of AGSA

There will be a need add to the traditional role of supporting, informing patients, families and the community about genetic disorders. AGSA will need to be kept up to date and well informed to technological advances.

Consider next the developments in genetics based on genomics.

Advocacy role – in the multifactorial disorders there are already comparable support bodies like JDRF with lots \$\$\$\$. Where does AGSA and comparable organisations sit vis-à-vis these organisations.

Future

We are still in the discovery era – genomics, proteomics metabolomics etc etc.

Problem - we know a lot - but what can we do about it?

New therapeutics are developing and the genetics of the future will focus more on outcomes than diagnosis – benefiting patients, families and the community in general.



**GENETIC AWARENESS WEEK LAUNCH
NAB, GEORGE STREET, SYDNEY
8TH AUGUST 2007.**



Dianne Petrie thanking Lynne Foxall



Laurie Taylor, Flavia Abbate, Peter Allison



Zoe Horton, Peter Allison



Kathy Fitzgerald announcing the Raffle Prize Winners - Val Thomas won a Dinner for Two at Bambini Trust Wine Room, Sydney and David Oliver won a bottle of Moet & Chandon Champagne. Congratulations!!!



Prof David Sillence



Dianne Petrie, Ellie Smith, Jenny Elliot



BIONEWS

'Reprinted with permission from BioNews (www.bionews.org.uk)"a free web-and-email-based news service on human genetics and assisted reproduction, published by the UK charity Progress Educational Trust.

GRANDFATHER TO DONATE SPERM FOR SON'S CHILD:

By Ailsa Taylor:

A 72-year-old grandfather will be allowed to act as a sperm donor for his infertile son and daughter-in-law, effectively making any resulting child his father's biological half brother. Although theoretically not illegal, according to the Human Fertility and Embryology Authority, is thought that this is the only time such a case has arisen in the UK.

The decision to allow the couple to use the grandfather as a sperm donor follows many months of consultation with independent bodies and ethics committees, says Kamal Ahuja, co-medical director of the London Women's Clinic where the couple are being cared for.

Ahuja believes that the couple's wish to have a child that is similar to their own identity is understandable. 'We have made the decision on the basis that the couple have special requirements in that the donor sperm is not acceptable to them', he told the Guardian. 'That applies to many, many groups of people - some religions [such as Islam] don't condone the use of donor sperm. In this particular case there was a mixture of reasons'.

However the national sperm donation programme accepts only men aged under 45, raising concerns over the increased risks of miscarriage and genetic mutation associated with the raised paternal age. Allan Pacey, of the British Fertility Society, told the Times: 'I am sure the couple will have been apprised of the risks, but in my view this is unwise. There is a very real possibility that this will not work, and the chances of miscarriage are also raised. The chances of a genetic defect or illness become greater too. You could say that if everybody is happy they should go ahead, but God forbid if there is a child born with a problem. It would be delicate to explain to that child that it might be the result of its grandfather's 72-year-old sperm'.

According to the BBC, the couple, who are in their 30's and have chosen

to remain anonymous, have not yet decided whether they will tell the child the true identity of his or her biological father, although the clinic is positively encouraging them to do so.

- The Guardian 6/10/2007 '72-year-old sperm donor to father his own grandchild'

- The Guardian:

<http://www.guardian.co.uk/science/2007/oct06/medicalscience/print> -

<http://news.bbc.co.uk/1/hi/health/7030267.stm>

- BBC News Online 5/10/2007 'Man donates sperm for grandchild'

POSITION VACANT at AGSA

Position Title: Project Officer

Part time – 24 hours per week

The Association of Genetic Support of Australasia

(AGSA) supports people affected directly or indirectly by genetic conditions. AGSA has the largest contact registry of over 750, mostly rare, genetic conditions plus 200 rare chromosome abnormalities. AGSA supports individuals and families through seminars, sibling workshops, facilitating contact with others with the same / similar condition and through telephone contact.

AGSA is seeking an enthusiastic and creative individual to facilitate their Filling the Void project. Filling the Void was established in 2005 through funding from NSW Health and aims to support individuals and families caring for someone with a genetic condition through the provision of the following:

- *rural and metropolitan seminars
- *rural and metropolitan sibling workshops
- *telegroup counselling
- *face to face counselling

Essential selection criteria:

Tertiary qualifications in social work, health sciences, science, nursing or counselling discipline.; training and experience in counselling, understanding of carer issues; excellent communication, interpersonal, computer and organisational skills; ability to work alone and as part of a team; current driver's license; must be able to undertake travel throughout NSW and some evening/weekend work.

The successful applicant will demonstrate a working understanding of the impact of living with genetic conditions on the individual, family and community. An understanding and commitment to equitable access to this program is essential.

A willingness and ability to promote this project through presentations to Genetic Counsellors, other relevant support services and at conferences is desirable.

A Working with Children's Check will be conducted on the preferred applicant. For a full position description and any enquiries contact Dianne Petrie on 9211 1462. Applications close on 30th October 2007 To AGSA, 66 Albion St, Surry Hills, NSW, 2010.



The Association of Genetic Support of Australasia Inc. (AGSA)

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Office Hours:

Mon – Wednesday 9 am – 5 pm

Thurs – Friday 10 am – 4 pm

Medical and Professional Advisory Board

Dr K. Barlow-Stewart PhD; BSc

Prof. D. Sillence MB BS; MD (Melb; FRACP;
FRCPA, FAFPHM

Prof B Wilcken MB;ChB;FRACP

Prof. R.Trent PhD; BSC (Med); MB BS (Syd;
BPhil (Oxon), FRACP; FRCPA.

Subscription Year 1st July - 30th June

ANNUAL SUBSCRIPTION

Individual \$24.00 incl.GST

Group/Organisation \$48.00 incl.GST

AGSA aims to:-

- * provide a contact point for families who are affected by genetic conditions so rare that they do not have their own support group.
- * facilitate access to individual support groups for those families with a particular genetic disorder.
- * 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.

** The views expressed in this Newsletter are not necessarily those of AGSA**



**AGSA'S SUPPORT GROUP & ORGANISATIONAL MEMBERS
as at January 2003**

Act Muscular Dystrophy Association Inc.
Androgen Insensitivity Assoc. Support Group of Australia
Alagille syndrome Support Group
Albino Support Group
Angelman syndrome Assoc. Inc.
Alzheimer's Assoc of Aust Inc.
A.P.I.A. (Aust.Primary Immune Deficiencies Assoc.)
Assoc. for Children With a Disability, Vic.
Assoc. for the Welfare of Child Health (AWCH)
AUSSIE FOLKS
Australian Addison's Disease Assoc. Inc.
Aust. Arthrogyrosis Group (TAAG) Inc.
Australian Assoc. for the Welfare of Child Health (AWCH)
Aust. CHARGE Association
Aust. Crohn's & Colitis Assoc.
Aust. Huntington's Disease Association (Qld) Inc.
Aust. Huntington's Disease Assoc. (NSW) Inc.
Aust. Speak East Assoc.
Australasian Tuberosus Sclerosis Society Inc.
Aust. Leukodystrophy Support Group
Aust. Society for Ectodermal Dysplasia
Autistic Assoc. of NSW
Batten's Disease Support & Research Assoc. Inc. (Australian Chapter)
Beckwith-Weidemann syndrome Support Group
Bunyip Special Needs Group Inc.
Cardiomyopathy Assoc of Aust. Ltd.
Centacare Early Intervention.
Centre for Developmental Disability Studies
Charcot Marie-Tooth Assoc. of Australia Inc.
Charcot Marie-Tooth Disease, USA
Child & Family Health Centre
Child Health Information Centre
Community Resource Team (Albury)
CONTACT A FAMILY U.K.
Cleft Pals, The Cleft Palate & Lip Society
CLIMB Children Living with Inherited Metabolic Diseases
Coeliac Society of NSW Inc.
Congenital Adrenal Hyperplasia Support Group
Cornelia de Lange syndrome Support Group
Cri du Chat syndrome Support group of Australia Inc.
CVS Support Group (WA)
Cystic Fibrosis Assoc of Qld Ltd.
Cystic Fibrosis Assoc. of Vic
Cystic Fibrosis New South Wales
Early Education Clinic, North Sydney
Early childhood Intervention Program
DIAL (Qld)
Donor Conception Support Group
Depressive & Manic Depressive Assoc.
Dystrophic Epidermolysis Bullosa Research Association (DEBRA) NSW. Inc.
Early Learning Tasmania
Ehlers-Danlos syndrome Support Group
Exceptional Parent (USA)
Fabry's Support Group Inc.
Family Advocacy
Family Planning Assoc.
Fragile X Assoc of Australia
Friedreich Ataxia Assoc of NSW
Gaucher Assoc. of Australia
Genetic Alliance (USA)
Genzyme Australia Pty. Ltd.
Genetic Interest Group (GIG) (UK)
I.D.E.A.S. Inc
Kidney Kids Support Group NZ
Klinefelter syndrome Support Group
Kurrajong Early Intervention
Haemochromatosis Society Inc.
Haemophilia Foundation NSW
Hereditary Cancer Registers (NSW Cancer Council)
Hereditary Haemorrhagic Telangiectasia
Hereditary Fructose Intolerance
Hunter Orthopaedia School
IDEAS Inc.
Kidney Kids of NZ Support Group
Maternity Alliance
NALAG
Leukodystrophy Foundation (USA)
Leigh's Disease Support Group
Lowe's syndrome Assoc. Inc. (USA)
Lower Nth Shore Community Support Team
Lupus Association of NSW Inc.
Lysosomal Diseases Australia
M.P.S. Society
Marfan syndrome Support Assoc. NSW
Marfan syndrome Assoc. Australia (S.A.Branch))
Meniere's (NSW) Support Group
Mental Illness Nervous Disorders Association
Metabolic Dietary Disorders Association (MDDA)
Mid North Coast Area Health Taree Genetics Service
Motor Neurone Disease Assoc. of NSW Inc.
Multiple Epiphyseal Dysplasia Assoc.
Muscular Dystrophy Assoc of NSW
Muscular Dystrophy Assoc (NZ) Inc.
National Council of Intellectual Disability
NCOSS (NSW Council of Social Services)
Neurofibromatosis Assoc.
Noonan syndrome Support Group
NSW Genetics Education Program
NSW Cancer Council
Osteopetrosis Support Group
Osteogenesis Imperfecta of Aust.
Parents Bereavement Support Group
Parent to Parent (NZ)
Pen-Parents of Aust. (ACT)
PKU Assoc of NSW
Polycystic Kidney Disease Association
Psoriasis Society
Pseudohypoparathyroidism Support Group
Pseudoxanthoma Elasticum Support Group
Prader-Willi syndrome Assoc. of NSW (Aust) Inc.
Pyruvate dehydrogenase deficiency.
Rare Chromosomes Disorders Support Group
Retina Australia (NSW) Inc.
Rett syndrome Assoc. of Aust.
Royal Blind Society of NSW
SAFDA (Support After Foetal Diagnosis of Abnormality)
SANDS
Short Statured People of Northern Qld
Short Statured People of Aust (NSW)
Short Statured People of Aust (Vic)
Short Statured People of Aust. (SA)
Spinal Muscular Atrophy
Schizophrenia Fellowship NZ
Smith Magenis syndrome Support Group Inc.
Spastic Society of Victoria
Spina Bifida Assoc. of NSW
Spina Bifida Assoc. of WA Inc.
Society of Ectodermal Dysplasia
SOFT Australia
Southern Child Care Support Program
Sotos syndrome Support Group
Steele Street Early Special Education Centre Devonport
St Paul's Special School
The Chromosome 18 Registry & Research Society
The Northcott Society
The Toybox Centre Inc.
Thalassaemia Society of NSW
Turner syndrome Assoc of Aust. Ltd. (QLD)
Turner syndrome Assoc of Aust. Ltd. (SA)
Turner syndrome Assoc. of Aust. Ltd. (NSW)
Uncontrolled Epilepsy Support Assoc (Vic)
United Leukodystrophy Foundation (USA)
Velo-Cardio-Facial syndrome Foundation of Australia.
Wellington Huntington's Disease Assoc. (Inc.) (NZ)
Western Institute for Self Help (W.I.S.H)
West syndrome Support Group
Wolf-Hirschhorn 4p- syndrome Support Group
Williams syndrome Association of Aust. Inc.

(NB: This list represents support groups and associations members only. In addition to this list of members AGSA has established a Contact Register over 550 genetic conditions representing families and individuals seeking contact.)

