



**THE ASSOCIATION  
OF GENETIC SUPPORT OF  
AUSTRALASIA INC.**

FUNDED BY THE NSW HEALTH DEPARTMENT

NEWSLETTER

**DECEMBER 2000 ISSUE 50**

**MISSION STATEMENT**

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

Reg Charity CFN15481 ABN 83 594 113 193

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**EDITORIAL**

The United Nation has proclaimed 2001 The International Year of the Volunteer. Just think of all the support group coordinators who have been doing such an excellent job unrecognized for many years. There are over 700 support groups fulfilling a very great need in the community. Writing newsletters, organizing conferences, picnic days and providing up to date information to members and professionals. This work is time consuming and often is done on a shoestring. Congratulations to all the committee members and founders of these much needed support group.

I would like to thank the support groups for including AGSA on their mailing list. It keeps AGSA up to date and provides a valuable reference point for information and referring callers.

AGSA is holding a planning day the first week in February wherein the meetings for the year will be planned. We have had many requests to hold a Triple X and a Double Y seminar. If you would like AGSA to assist with your group in organizing a get together with a guest speaker please let me know. AGSA runs four support group seminars a year plus Genetic Awareness Week.

I hope 2001 is kind to you  
Best wishes,

Dianne Petric

## CONTACT CORNER

*AGSA will publish requests for contacts 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.*

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### FAMILIAL HIBERNIAN FEVER

A lady with two affected family members would desperately like contact with others who have this condition. Contact AGSA for details.

### PROTEUS SYDNROME

A parent of a two half year old boy would like contact with other people who have had experience with this condition. Contact AGSA for details.

## SUPPORT GROUP NEWS

*This section highlights some of the support groups and what is currently happening within the groups. The information is taken from their newsletters and maybe of benefit to our members. Contact AGSA for more details.*

### Cardiomyopathy Association of Australia - Winter, Volume 24 2000

The association has now incorporated. They have several tapes for loan to members eg diagnosis, exercise, overview of cardiomyopathy, medical & surgical technologies. They have found that members have had difficulty in finding Insurance Companies to cover them for travel insurance and have information regarding this They offer a phone-link service.

### Sygnnet - Syndrome Without A Name -

The UK based group have highlighted in their newsletter about sblings and also some book reviews.

'Workshops for Siblings of Children with Special Needs' - authors Donald J. Meyer and Patricia F. Vadasay. Jessica Kingsley Publishers 020-7833-2307

'Tom, A Gift In Disguise' - By Henrietta Rose, Findhorn Press 01309-690582. Book is written by a mother of an undiagnosed child.

'Bringing Up a Challenging Child At Home - When Love Is Not Enough' - by Jane Gregory, Jessica Kingsley Publishers 020-7833-2307.

'David and the Timid Dinosaur', 'When Your Beautiful Child's Not Normal', 'David And The Tooth Fairy', 'David Needs The Toilet' - author and illustrated by Wendy Webb who was inspired by her children with Autistic Spectrum/Aspergers Syndrome.

### Turner Syndrome Association of Australia Ltd - June 2000

Have reprinted an article 'Ten Tips For Finding A Medical Professional For Your Child' by Jan Hunt, from the Natural Child Project website at [www.naturalchild.org](http://www.naturalchild.org)

### IDEAS - Information on Disability - Equipment Access Services- December 99 /August 2000

As the name implies, they have information on equipment for all ages and disabilities. They have written a short article in their newsletter on the NSW Apprenticeship Program for People with Disabilities, & Are you entitled to Purchase a Car GST Free?

### NSW Meniere's Support Group Inc. - June 2000

The 'Balancer' newsletter has reiterated the importance of a Medical Alert Card for their members. This is also of necessity for anyone with any ongoing medical illness. Salt Free Worcestershire Sauce recipes. For allergy sufferers, special baked bread is available from Pav's Products - [www.pavsallergybakery.com.au](http://www.pavsallergybakery.com.au) or (02) 9829-7811

**Congratulations to Genetic Support Network of Victoria** who have secured increased funding to employ two Genetic Support Coordinators on a job-share basis. Contact Caroline Bowditch on (03)8341 6315

**News Flash** The National Library of Australia have been assessing online publications since 1996 and identifying those that they consider to have "national significance" and that are recognized as being of "lasting cultural value". Those identified as meeting with their criteria are then asked for permission to be included in

their archives so that "Australians have access to their documentary heritage now and in the future".

The AIS Support Group Australia Newsletter have been invited to be included in their archives.

Congratulations.

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**Government Leaves People With Disability Fearful**

**Chris Evans – Acting Shadow Minister for Family and Community Services**

**Media Statement 15 December 2000**

Labor offices have been inundated with calls from people with disability in the last 24 hours all deeply concerned about what the Government's welfare reform agenda will mean for them." The Government has created great distress for people with disability by failing to guarantee that they will not be ruled ineligible for the Disability Support Pension" said Senator Chris Evans. Acting Shadow Minister for Family and Community Services. "To leave a vulnerable group in our community feeling scared and uncertain about the future is insensitive, unnecessary and unfair". "The Minister has left the door open to shift people with disability from DSP to Newstart through a "more robust and independent' assessment process in which government contracted medical experts will assess eligibility. "Persons ruled ineligible for DSP under the tighter guidelines will face reduced benefits, tougher activity tests, and harsh penalties if they are unable to meet their mutual obligation requirements. "The explosion in penalties applied to jobseekers – an increase of 250 per cent in the last three years –has already caused serious problems for people with intellectual disability. People who cannot understand Centrelink letters or who miss appointments because of the episodic nature of mental illness are being caught in the Government's breaching web. "the Government needs to provide additional support to people with disability not additional sanctions. "the Minister's statement yesterday failed to acknowledge that the majority of people with disability desperately want to work. They want real jobs but employment opportunities are thing on the ground. "As the McClure report pointed out, business has provided few participation opportunities for people with disability. But before the Government shifts responsibility to business they should clean up their own act. "Under the Howard Government more than

2000 people with disability have lost positions in the Commonwealth Public Service. "Tow Centrelink employees in Western Australian have just been made redundant because their disability makes them unsuitable to perform tasks associated with customer service a and multi-skilling. Despite combined service of 21 years they were not offered either redeployment or retraining." "People with disability have a right to be skeptical about the Government's unfounded commitment to additional employment services. It is skepticism based on evidence. They also have a right to know what the future holds. "Labor calls on the Prime Minister to assure people with a disability that the Government's welfare reform agenda will leave no-one worse off. Failure to act now will leave people with a disability facing a fearful and uncertain Christmas."

Authorised by Geoff Walsh 19 National Circuit, Barton Act 2600.

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*The announcement was made public on Friday 10th November and filmed by Channel Nine News. The story went to air Tuesday 14th November 2000 Channel Nine News.*

**Media Release.**

**"I decided to do something about this...."**

Dr Michael Wooldridge, local MP and Health Minister, has joined with Kerri Carboon of Montrose, the President of the Metabolic Dietary Disorders Association to announce that the Federal Government will provide special support to the suffers of Inborn Errors of Protein Metabolism and their families.

Inborn Errors of Protein Metabolism are relatively rare disorders, with less than 1,000 sufferers Australia wide. Phenylketonuria (PKU) is the most common of these, with approximately 750 people affected.

"The sufferers cannot eat the foods the rest of us take for granted.

"Their special diets are mostly made up of imported foods that are also very expensive and they must maintain the diet to avoid permanent brain damage.

"Kerri and her husband Neil came to see me as their local MP. I was deeply touched when they told me about their own experiences as parents of two children with PKU," Dr Wooldridge said.

Mrs Carboon has been actively involved in establishing and building on an existing Australia-wide support network for families with members who suffer from these dietary disorders.

“During the meeting with Neil and Kerri I decided to do something about this.

“As a local MP, you often confront issues over which you have very little control but as the Health Minister this was an issue that I could actually do something about,” Dr Wooldridge said.

Dr Wooldridge announced that the Federal Government will provide support of \$2,400 a year for every person (child or adult).

“This means that families who have children with one of these disorders will benefit from the support and the child will continue to receive the support when they reach adulthood.

“This is very important because it gives them certainty. They should not have to pay so much just to eat.

“Kerrie has worked very hard on this and I am very glad that I, as her local MP, have been able to do something that will actually change her life and those of other families for the better,” Dr Wooldridge said.

## PROFILE PKU

Facts about Phenylketonuria (PKU) and other inborn errors of protein metabolism.

- PKU is an inherited inability to metabolise phenylalanine, an amino acid component of protein common in many foods.
- If phenylalanine is consumed by people with PKU it essentially acts as a toxin and in a short period of time can lead to mental retardation (particularly in babies and young children) and neurological problems such as convulsions.
- PKU can be managed by a diet of special low protein, high-fat foods, and phenylalanine free, protein-substitute formula.
- there are a number of other “inborn errors of protein metabolism” - much less prevalent than PKU, the most common - which also require consumption of these special foods.

- The protein-substitute formulas are available on the PBS but the modified foods (bread, pasta, flour, biscuits) have to be purchased commercially - most are imported.
- It is estimated that the cost of these foods can be 5-10 times the cost of normal foods and place serious financial hardship on families - particularly those with more than one child that suffers from the condition.
- It is estimated that there are approximately 750 people (children and adults) in Australia that have PKU and up to another 250 that have other protein metabolism disorders.
- Accounting for the high costs of those special foods, the Commonwealth is proposing to provide an annual grant of \$2,4000 in each patient that would cover about half of the annual costs of these foods.

### Inborn errors of protein metabolism

<u>Disorder</u>	<u>Incidence</u>
Phenylketonuria	1:14,000
Maple syrup urine disease	1:120,000-500,000
Homocystinuria	1: 50,000-100,000
Urea cycle disorders	unknown
Propionic acidaemia	unknown
Methylmalonic acidaemia	unknown
Tyrosinaemia 1 and 2	unknown
Lysinuric Protein intolerance	unknown
Arginase deficiency	unknown
Glutaric acidaemia	unknown
Hyperornithinaemia	unknown

*This list is not absolute and new errors may be diagnosed in the future. Any such errors that are determined by a specialist to require the special foods will be covered by the grant.*

### **Metabolic Dietary Disorders Association (MDDA)**

Kerri Carboon (02) 9728 5510

Natalie Wiggins (02) 9850 7484

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## CONFERENCES

**Women with Disability Australia (WWDA) are holding** a forum to talk about sterilization of women with disabilities. This is the first time women with disabilities will gather to discuss their experiences and rights in this matter. The national forum will take place in Sydney on Thursday 15th February 2001.

Remember and look forward

An international conference in Sydney on 16th-17th February 2001 will mark the twentieth anniversary of the International Year of the Disabled Person.

'Disability with attitude: critical issues 20 years after International Year of the Disabled Person' will examine progress, celebrate achievements and address critical disability issues facing Australian society.

Keynote speakers include Professor Adrienne Asch, Dr Tom Shakespeare, Don Grimes, Phillip French and Helen Meekosha.

Contact: (02) 9385 1870 Fax (02) 9662 8991

## IS GENETIC TECHNOLOGY GETTING OUT OF HAND? – ISSUES OF GENETIC SCREENING.

**2<sup>nd</sup> March 2001**

**10 a.m. – 4.30 p.m.**

**KEYNOTE SPEAKER Tom Shakespeare**

**Venue: Centre for Ministry,**

**16 Mason's Drive,**

**North Parramatta**

For Contact and Registration: ELM Centre, 02 9683 3655

Email [elm@nsw.uc.org.au](mailto:elm@nsw.uc.org.au)

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**THE CHROMOSOME 18 REGISTRY & RESEARCH SOCIETY (AUSTRALIA)** is currently organizing a conference for the latter half of 2001 in Sydney. For further enquiries please contact (02) 9580-5707

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## THE BATTEN DISEASE CONFERENCE 2001 IN AUSTRALIA

**8.30 am – 5pm Saturday 21<sup>st</sup> March and**

**Sunday 1st April 2001**

**"Dockside" Cockle Bay side of darling Harbour, Sydney**

**Morning Tea, Lunch & Afternoon Tea – price to be advised.**

**Accommodation:** The Parkroyal, Darling Harbour holds a group book – information to be supplied.

The Australian Chapter of Batten Disease Support & Research Association aims to provide families and friends affected by this disease with information, education and social interaction in various aspects of Batten Disease.

Guest speakers to include worldwide researchers and medical professionals.

**Contacts: Vanessa Anderson Ph: (02) 4334 5785 Beth Dowry Ph: (02) 4261 1655**

**Batten Disease Support & Research Association**  
[www.bdsr.org](http://www.bdsr.org)

5 Oakshurst Close, Dapto NSW 2530 Register Charity No CFN 14877 ABN 56056118994 Email: [gvicando@ozemail.com.au](mailto:gvicando@ozemail.com.au)

**We are happy to announce the 2001 Battens Conference will be taking place end of March at Darling Harbour, Sydney.**

The mission of this Conference is to provide families and friends affected by Battens Disease or alike with the opportunity together at a central location in a friendly environment. To provide time for social interaction and information, training, and education in various aspects of this disease.

Our BDSRA USA groups have organized for worldwide researchers to share their knowledge updating us on research over the two day Conference duration.

**\*\*We would like to hear from any persons (family, friends, medical professionals) interested in attending, to assist us with booking numbers at the venue.**

**\*\* Sponsors and/or donators are sought to assist families to travel from distances around Australia and New Zealand. Financial assistance will aid nursing and carer requirements.**

**\*\*Medical and field specialist guests will be approached to speak on issues that are part and parcel of the disorders.**

**\*\*If you assist caring for the affected kiddies and their siblings please phone Vanessa on (02) 4333 5785.**



## **AGSA SUPPORT GROUP MEMBERS as at January 2001**

*Act Muscular Dystrophy Association Inc.*  
*Androgen Insensitivity Assoc. Support Group of Australia*  
*Alagille Syndrome Support Group*  
*Albino Support Group*  
*Alliance of Genetic Support Group, U.S.A.*  
*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*  
*Aust. Arthrogryposis Group (TAAG) Inc.*  
*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 Tuberous Sclerosis Society Inc.*  
*Aust. Leukodystrophy Support Group*  
*Aust. Society for Ectodermal Dysplasia*  
*Autistic Assoc. of NSW*  
*Batten's Disease Support & Research Foundation*  
*Beckwith-Weidemann Syndrome Support Group*  
*Cardiomyopathy Assoc of Aust. Inc*  
*Centacare Early Intervention.*  
*Charcot Marie-Tooth Assoc. of Aust Inc.*  
*Charcot Marie-Tooth Disease, USA*  
*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*  
*Comelia de Lange Syndrome Support Group*  
*Cri du Chat Syndrome Support group of Australia*  
*CVS Support Group (WA)*  
*Cystic Fibrosis Assoc of Qld Ltd.*  
*Cystic Fibrosis Assoc. of Vic*  
*Cystic Fibrosis Foundation, North Ryde.*  
*Early Education Clinic, North Sydney*  
*DIAL (Qld)*  
*Donor Conception Support Group*  
*Depressive & Manic Depressive Assoc.*  
*Dystrophic Epidermolysis Bullosa Research Association (DEBRA) NSW. Inc.*  
*Ehlers-Danlos Syndrome Support Group*  
*Exceptional Parent (USA)*  
*Fabry's Support Group Inc. (Vic)*  
*Family Advocacy*  
*Family Planning Assoc.*  
*FAP Register (NSW Cancer Council)*  
*Fragile X Assoc of Australia*  
*Friedreich Ataxia Assoc of NSW*  
*Gaucher Assoc. of Australia*  
*Genzyme Australia Pty. Ltd.*  
*Genetic Interest Group (GIG) (UK)*  
*I.D.E.A.S. Inc*  
*Klinefelter Syndrome Support Group*  
*Kurrajong Early Intervention*  
*Haemophilia Foundation NSW*  
*Hereditary Haemorrhagic Telangiectasia*  
*Hereditary Fructose Intolerance*  
*Hunter Orthopaedia School*  
*Huntingtons Disease Assoc. (NSW)*  
*Huntingtons Disease Assoc. (QLD)*  
*IDEAS Onc.*  
*Maternity Alliance*

## **NALAG**

*Leukodystrophy Foundation (USA)*  
*Leighs Disease Support Group*  
*Lowe's Syndrome Assoc. Inc. (USA)*  
*Lower Nth Shore Community Support Team*  
*Lysosomal Diseases Australia*  
*M.P.S. Society*  
*Marfan Syndrome Support Assoc. NSW*  
*Marfan Syndrome Assoc. Australia (S.A. Branch)*  
*Meniere's (NSW) Support Group*  
*Metabolic Dietary Disorders Association (MDDA)*  
*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*  
*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*  
*Psoriasis Society*  
*Pseudohypoparathyroidism Support Group*  
*Pseudoxanthoma Elasticum Support Group*  
*Prader-Willi Syndrome Assoc*  
*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*  
*The Chromosome 18 Registry & Research Society*  
*The Northcott Society*  
*Thalassaemia Society of NSW*  
*Turner Syndrome Assoc of Aust. Ltd. (QLD)*  
*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)*  
*West Syndrome Support Group*  
*Wolf-Hirschhorn 4p- Syndrome Support Group*  
*Williams Syndrome Association of Aust. Inc.*

*(NB: This list represents support groups and associations only. In addition to this list of members AGSA has established a Contact Register over 500 genetic conditions representing families and individuals seeking contact.)*

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

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**Peer Support/Information  
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**Dianne Petrie**

Office Hours: 10.00 am - 3.00 pm  
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Closed Friday

**Medical and Professional Advisory  
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FRACP; FRCPA.

**ANNUAL SUBSCRIPTION**

Individual	\$22.00
Group/Organisation	\$44.00

*Subscription Year 1st July - 30th June*

**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 \**