



# THE ASSOCIATION OF GENETIC SUPPORT OF AUSTRALASIA INC.

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NEWSLETTER

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## 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

AGSA is presently up-dating its website and we would like to extend an invitation to our members to be on it. If you wish to advertise your support group, latest research news or a proposed conference, please fax or email me the details or give AGSA permission to insert your support group contact details. This information will be placed in the News section or support group section of the website. For the month of June 2001 AGSA's website received 7,821 hits and has an average monthly total of 6,995 hits.

AGSA's AGM will be held on Sunday 21<sup>st</sup> October 2001 at 10.00 a.m. Dr Kris Barlow Stewart will be our guest speaker. All are welcome especially those who wish to join the committee.

Up coming meetings are Breast Cancer Testing Information Day on 7<sup>th</sup> October, Triple X syndrome on Sunday 14<sup>th</sup> October and The Rare Chromosome Disorders Support Group Seminar on Sunday 28<sup>th</sup> October 2001. All these meetings will be held at AGSA 66 Albion Street Surry Hills.

I am pleased to announce AGSA's Charity Status has been renewed and is valid till 2006.

I recently attended the Human Genetics Scientific Conference in Cairns, which resulted in some media coverage about AGSA on Brisbane radio. The conference was very informative and provided a good opportunity to catch up with people and learn about the latest research.

I look forward to seeing you at the AGM.  
Best wishes

**DIANNE PETRIE**

## **MEMBERSHIP FEES – AGSA IS NOT REGISTERED FOR GST**

AGSA's Membership is due as of 30<sup>th</sup> June 2001 and it appears some organisations are confused regarding GST.

A not-for profit organization is required to register for GST if its annual sales from all sources is \$100,000 or more. By being registered for GST purposes, an organization is required to pay to the ATO GST on their sales, and are able to claim tax input credits. Such organizations would more than likely include GST on their sales.

A not-for-profit organization does not have to register for GST purposes if its annual sales from all sources are under \$100,000 e.g. AGSA Organisations that do not register are not required to charge the GST on their sales, and are unable to claim any input tax credit on purchases. If such organizations choose to register, they would more than likely include GST on sales and pay to the ATO one eleventh of the sales less any input tax credits.

AGSA is not registered for GST therefore our membership does not include GST and payment is a straightforward \$22 for individual or \$44 for an organization without having to claim or pay back GST.

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## **VALE**

**Vaughan Glasson (The Sojourner) born at Goroka, Papua New Guinea 15<sup>th</sup> June 1975 to 31<sup>st</sup> August 2001 (26 years of age).** *Vaughan was an active supporter of AGSA for many years and we wish to acknowledge his support, fighting spirit and friendship by sharing with you his journey.*

Vaughan started travelling at 10 days of age on a flight from Goroka to return to his home on a coffee plantation in the Banz area, where he spent his babyhood. Before he was three, he set off on a trip around the world with his older brother and parents. He was too young to

remember the 40 odd countries he visited, but he enjoyed them.

In Australia he was diagnosed with MPS2 (Hunter syndrome) on Friday 13<sup>th</sup> about the time of his 4<sup>th</sup> birthday.

Regular speech therapy was part of his life in preschool and infants school. There were 200 applications for six places in the Opportunity Language Class at Gordon West Public School. Vaughan was lucky to secure one of those places for the ensuing two years. Despite deafness and with the help of an itinerate teacher for the deaf, Vaughan did well at Primary School. He was the inaugural winner of the Hugh Barlett Shield – awarded for achievement and perseverance over difficulties. (\*He wore aids in both ears).

As a youngster he rode his bike and skateboard, climbed trees and played games along with the neighborhood lads.

He attended Shore School at North Sydney where he successfully sat for his Higher School Certificate. By high school he needed to wear glasses, he could not play contact sports; his stiff joints and breathlessness were becoming real problems. He was plagued by frequent ear infections, double vision, colds and bronchitis. Despite these limitations he aqua boarded, went snow ski-ing, played soccer and snow boarded. He excelled at snooker and pool. He loved board and card games. He memorized the cards played in each game. Every one wanted to be his partner!

After school Vaughan completed an 18 months Bookkeeping Certificate course and worked part-time for the CRS for some years and was still working one day per week bookkeeping at Pepe's Restaurant, Balmain.

He had many operations – ear grommets, 5 hernia repairs, carpal tunnel release of both wrists. The C-PAP machine was part of his life every night for the last nine years. Vaughan travelled (3 times on his own) to every Australian state and to New Zealand twice – lugging his C-PAP machine with him.

From 18 years he participated on a large scale with the MPS Society. He was the official video

recorder at the MPS conference, Wollongong in 1991. This was where parents and professionals first realized that they had a rare opportunity to gain an insight into MPS because Vaughan was willing to share his thoughts and feelings about MPS with them. At the International MPS Conference, in May 1996, Vaughan was one of a brave group of young MPS individuals who shared what it meant to them to be affected by MPS.

His computer gave him the power to reach out to others via email. He was the assistant editor of the MPS newsletter "The Linking Hand" and his major effort for the society was to compile, with the help from others, a booklet entitled "Towards greater independence – guide for adults with mild MPS". He was a Director of the MPS Society at the time of his death.

Anyone who had had the opportunity to meet Vaughan would know he had opinions on most subjects and when he felt strongly enough about something, he did something about it. His comments were included once on the "60 Minutes", and recently he was interviewed on living with MPS by Madeline Randall on ABC regional radio.

Just before his death he had completed, published and distributed his book on the Glasson Family History entitled "The Sojourners" (214 pages). This was the effort of five years research.

*Is it any wonder, a very diverse group of people came from all over NSW and interstate to say goodbye. He was piped to and from the church by his cousin, and given a Police escort by his brother's mates from the church to the Crematorium – Vaughan would have liked that. Fond memories. Our wishes to Trev and Isabel, Brennan and Jackie.*



## Support Group News as at 12<sup>th</sup> August 2001

**IDEAS**, Information on Disability – Equipment Access Services, June/July 2001 Newsletter - [www.ideas.org.au](http://www.ideas.org.au). Their newsletter features 'The Access File'. The current article features accessible attractions and venues in Canberra, e.g. Cockington Green, Questacon. For more information ring Kellie on 1800 029 904

**Donor Conception Support Group** – July/August 2001 Newsletter. A new book 'Is My Baby All Right' by Christine, Gosden, Nicolaides & Whilling, has been added to the National Library.

**Australian Addison's Disease Assoc Inc** – July 2001 Newsletter. Website: [www.addisons.org.au](http://www.addisons.org.au). University of Sydney will be part of an international study looking into auto-immune endocrine disease relationships. For more information contact Professor Bruce Robinson, Royal North Shore Hospital, Endocrinology Department. Phone: (02) 9926-7111. The Better Health Channel have articles on various conditions and diseases – website: [www.betterhealth.vic.gov.au/bhcv2/bhcsite.nsf](http://www.betterhealth.vic.gov.au/bhcv2/bhcsite.nsf) or Phone: (03) 9616-2845

**SNAP**, Special Needs Advocate For Parents. Spring 2001 Newsletter. USA based, the website is: [www.snapinfo.org](http://www.snapinfo.org). They have advertised a website for parents wanting a personalised joyful memory book designed especially for a child with special needs. There are 4 books available – [www.heartfeltpress.net](http://www.heartfeltpress.net)

**Turner Syndrome Assoc. of Australia** – June 2001 Newsletter – One of the articles highlighted in this issue is an excerpt and adaptation from the Ark Foundation (USA) called 'Spatial Relations and Learning' by Stockdale and Possin, The Website is: [www.newhorizons.org/spneeds\\_arkspatial.html](http://www.newhorizons.org/spneeds_arkspatial.html)

**In Touch, Muscular Dystrophy Association of New Zealand**. June 2001 newsletter. Website: [www.mda.org.nz](http://www.mda.org.nz)

**For information about the New Zealand Guillain Barre Syndrome Support Group**, Contact: (07) 862-6438 or (06) 751-1014

**The South Easterly, South East Health Newsletter**, June 2001. Parents in South Eastern Sydney can now benefit from the availability of Triple P, the Positive Parenting Programme. The Programme has been implemented by Area Mental Health, aims to provide parents with management techniques to reduce child misbehaviour. For more information contact: Karen Myors (02) 9350-2501.

**On the Beam, Lowe Syndrome Association, USA,** Summer 2001 – Website: [www.lowesyndrome.org](http://www.lowesyndrome.org). The organisation has begun to publish a new newsletter ‘Going On: Living with the Loss of a Loved One With Lowe Syndrome’ for families and friends who have experienced a loss of a child with Lowe syndrome. For more information contact: [info@lowesyndrome.org](mailto:info@lowesyndrome.org). LSA families in the USA and Canada may register with Brain & Tissue Bank for Developmental Disorders at the University of Maryland - for more information and the range of conditions already registered see their website [www.btbankfamily.org](http://www.btbankfamily.org).

**Centre for Developmental Disability Studies,** April 2001 newsletter. Website: [www.cdds.med.usyd.edu.au/](http://www.cdds.med.usyd.edu.au/) or Phone: (02) 8878-0500

**FPA Healthrites.** For a catalogue of FPA Health publications, available books, videos and information sheets on a wide of subjects which include disability, sexuality, parenting, counseling etc, contact customer service by phone: (02) 8752-4307, (02) 9716-6099 or see their website: [www.fpahealth.org.au](http://www.fpahealth.org.au).

## 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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### WAGR SYNDROME

A family in NSW requests contact with others.

### NEONATAL BARTTERS SYNDROME

A family would like contact with others.

### PEUTZ -JEGHER SYNDROME

AGSA has two families with a history of this condition who would like contact with others.

### ASPERGER SYDNROME

A few requests for contacts with others – ages 45 12 and 4 years old. Contact AGSA for details.

### SYNDROME X HYPERINSULIN ANAEMIA

Contact requested for an adult with this condition.

### MOSAIC DOWN SYNDROME SUPPORT GROUP

If you know of any families who would be interested please contact AGSA for details.

### DISABILITY NEWS

Microsoft has put out a range of Disability Enabled Software that helps people with disabilities use computers, here’s what Microsoft have to say about Microsoft Accessibility.

“Accessible computers and software programs can make it possible for everyone, including people with varying degrees of disabilities, to use these technologies successfully in work, education and recreation.



***FROG: I hope you like AGSA’s mascot. Why choose a frog? Frogs are vital for the ecology and many are very rare and need to be protected. The frog serves to remind you what AGSA represents, a caring supportive and informative organization which has been “Making the right connections since 1988”.***

## PROFILE

## A - Z GENETIC

**CONDITIONS**

*It is the intention of AGSA to profile, in each issue, a particular Support Group/Disorder, thus increasing awareness within our membership of the range of genetic conditions. Also it hopes that where overlaps occur in conditions, support Groups may liaise with each other and thus gain a broader understanding of facilities, aids, etc. that may be of value to your individual membership.*

*Please ensure that all support group information is recent and reliable. It is of paramount importance that you let us know your group is "Alive and Well" and happy to take referrals.*

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*Marlene Brightwell originally formed the Rare Chromosome Disorder Support Group of Australasia with some assistance from AGSA. However, as you may or may not be aware, Jeanine Thornton took over the running of the group when Janine Cody in America formally asked Marlene, to set up a sub branch of the The Chromosome 18 Register & Research Society in Australia. When Jeanine found she was unable to run the group the rare chromosome register was handed back to AGSA.*

*This is a very complex group and AGSA receives enquiries from all over the world from families trying to find another family with the same or similar condition. Families are very isolated and there is little or no information available for them.*

*AGSA is holding a Rare Chromosome Disorder Seminar on 28<sup>th</sup> October 2001. It is anticipated families will travel from inter-state for this meeting. Please circulate the enclosed flyer to families who you think would be interested in attending.*

### **RARE CHROMOSOME DISORDER SUPPORT GROUP OF AUSTRALASIA**

What is classed as a rare chromosome disorder?  
For many of us, we can only try to relate to a

genetic condition if a specific syndrome name is attached to it. This is where the Rare Chromosome Disorder Support Group differs to other genetic support groups that have been established. Over 100 families whose child has been diagnosed with a rare chromosomal abnormality have contacted us. Each one of the affected individuals is classed as rare. Nearly every individual has a different karyotype i.e. chromosomal constitution of the person. These congenital abnormalities do not fall into an easily recognised pattern, therefore are not regarded as an established "syndrome" - a syndrome being a collection of characteristics, which help characterise a genetic abnormality. Some of our children and individuals are considered almost unique in the world.

Medically, individuals vary from very mild to severe. All seem to be affected by their rare genetic condition in some way, be it, physical features, intellectual delay, speech delay, delay in gross and fine motor skills and a range of medical problems. The majority having multiple problems.

Families, individuals, and many health workers that contact us for the first time are unaware of the diverse range of disorders that encompass this group. Parents and carers need to be educated with the new cytogenetic terms they are met with, to gain an understanding of their own and other disorders in the group e.g. deletions inversions, marker, breakpoints, tetrasomy, translocations, mosaic, karyotype, p arm, q arm, ring and the list goes on.

In 1994, AGSA held two meetings for families to establish the Rare Chromosome Disorder Support Group. Firstly to collectively give a name to this group of disorders and secondly for families to be educated and to foster friendships. The group met on several occasions and in 1997 gathered formally at our Conference at the AGSA office. Dr Ellie Smith, Cytogeneticist, New Children's Hospital, Westmead and Bronwyn Butler from NSW Genetics Education Programs being our medical speakers. All parents on our Contact Register hope to contact other families whose child has the same or similar disorder as

theirs, to seek and exchange information, to speak to others who have been through the various stages of diagnosis, assessments, loss and grief. For many parents they know that it may be a long wait before they meet or are contacted by another family with the same or similar condition as their child.

Within this group, individual members have set up their own support networks - Ring 22 Support Group, S.O.F.T. (Aust), Chromosome 9 Support Group, XYY Syndrome Support Group and the Chromosome 18 Registry & Research Society (Aust. Region). Contact the AGSA office with your enquiries about these groups.

These are some genetic disorders that individuals have enquired about;

Chromosome 1 duplication

Ch2q 31 del

Ch2q-

Partial Trisomy 2p

Ch 2 inversion

Ch 2q(21-34 duplication

Ch 3 deletion

Ch 4p-, Ch 22q11 deletion

Ch 4q+

Ch 4p, duplication

Ch 4q-(q34-35)

Ch 5q partial duplication/unbalanced translocation

Extra material on 5q

Ch 6q+

Ch 6q-

Ch 7p-

Ch 7q

Extra material Ch 8, 90% mosaic

Ch 80-(23.1)

Tetrasomy 9p mosaic, isochromosome 9

Ch 9p-

Extra material on Ch 9

Ch 10p-

Ch 10q-

Ch 10p-

Ch 12p-

Ch 13q

Ch 13q-, marker

M(UPD)14 - Maternal Uni-parental Disomy

Inversion dup.15

Ch 15 marker, XYY syndrome

Ch 15q-(13q,14q)

Ch 17p duplication

Ch 18q-(q21)

Ch 18q-(21.2)

Ch 18q-(21.3)

Ch 18q-(q22)

Ch 18q-(q23)

Ch 18p-

Paracentric Inversion 18

Tetrasomy 18p

Ring 18, mosaic

Inversion 18

Ch 19q+(13.3-13.4)

Extra material on Ch 20

Ch 21 -

Ch 21q-(q22.3)

Ring 21

Ring 22 mosaic

Extra band on Ch X

XYY syndrome

### **Trisomy**

Trisomy 5q (translocation Ch 5 & 11)

Trisomy 8p

Trisomy 8

Trisomy 8, Unbalanced translocation Ch 8 & 13

Trisomy 9p

Trisomy 13

Partial Trisomy Ch11q

Trisomy 10q attached to ch15

Trisomy 15q23, unbalanced translocation Ch 5

Trisomy 17q25, duplication Ch 17q

Trisomy 18

Trisomy 18q

Partial Trisomy 18q

Trisomy 18p

Partial Trisomy 19q

Trisomy 20, mosaic

### **The following are unbalanced translocations:**

Ch 3 & 5

Ch 4 & 6

Ch 3 & 5

on Ch 4q

Ch 5 & 11q- (Jacobsen's syndrome)

Trisomy 5q (Ch 5 & 11)

Ch6+,18p-

Ch 6 & 12

Ch 9 & 14  
Ch10 deletion, Ch 19  
Ch 14 & Ch 20  
Ch 18p- Ch 15p+  
Ch 18p+ (p11.2) & 18p- (p21.2)  
Ch 18q- & Ch 17q+  
Ch 19p- & Ch 11

## CONFERENCES

### **The Chromosome 18 Registry & Research Society (Australia) 1<sup>st</sup> National Conference – 22<sup>nd</sup>-23<sup>rd</sup> September 2001 at Cronulla Sharks Leagues Club, Cronulla, NSW.**

Two speakers from San Antonio, Texas, USA, have been invited to educate families and up-date families on Chromosome 18 research. They are Janine Cody, who is the Founder and President of the Society & Dr Dan Hale, Endocrinologist. They will also be speaking at Sydney Children's Hospital, Randwick and The Children's Hospital, Westmead on Chromosome 18 disorders and growth.

For more information regarding the conference or speakers, please ring (02) 9580-5707.

### **TURNER SYNDROME ASSOCIATION OF AUSTRALIA LTD NATIONAL CONFERENCE - PERTH 6th – 7th October, 2001 CWA, 1174 Hay Street, West Perth**

#### **Speakers -**

Dr Geoff Byrne & Dr Fiona Frazer - Endocrinologists

Dr Andrew Bulloch - Cardiologist

Mr Max Keyt - In Vitro Fertilization

Ms Katrina Berkov - Motivational Speaker

Plus Perspectives Panel, Discussion Groups and Workshops

#### **For more information contact:**

Mrs Glenn Fisher - National President

Email: <turnersyn@netpro.net.au>

Telephone: (02) 9452 4196 Fax: (02) 9975 4037

**Australian Spina Bifida & Hydrocephalus Association National Conference. 6<sup>th</sup> – 8<sup>th</sup> September 2001 in Adelaide.** For more information contact (08) 8443-5220 or email [info@spinabifida.asn.au](mailto:info@spinabifida.asn.au)

**ARATA 2001.** Practical magic... Putting Theory into Practice. What is the real magic of assistive technology? To be held on 2<sup>nd</sup> - 4<sup>th</sup> October 2001 in Brisbane, QLD. Contact (07) 3858-5530 or email [arata2001@im.com.au](mailto:arata2001@im.com.au)

**Dealing with the Grief and Loss Process: A Workshop for Individuals/Family Members/Caregivers Living With The Effects of a Neuromuscular Condition – 15<sup>th</sup> September - Riccarton, Christchurch, New Zealand.** Contact: Muscular Dystrophy Association. PO Box 16-238 Sandringham, Auckland New Zealand. Phone/Fax: (09) 815-7260. Email: [director@mda.org.nz](mailto:director@mda.org.nz)

**Australian Huntington's Disease (QLD) Inc.** Dinner and Seminar – Friday 28<sup>th</sup> September 2001 at Education Centre, Princess Alexandra Hospital, QLD. For program and registration form contact: Phone: (07) 3391-8833 or email: [gahda@logicworld.com.au](mailto:gahda@logicworld.com.au)

**Climb Conference, Children Living with Inherited Metabolic Diseases – 21<sup>st</sup>-23 September 2001, Sheffield, UK.** Email: [info@climb.org.uk](mailto:info@climb.org.uk)

**27<sup>th</sup>-29<sup>th</sup> September 2001 Positive Steps Forward – National Biennial Autism Conference, Adelaide, SA.** Hosted by Autism Council of Australia. For further information, registration details and call for abstracts contact: Conference Secretariat, Plevin and Associates Pty Ltd. Ph: (08) 8379 8222: Fax (08) 8379 8177 Email [plevin@camtech.net.au](mailto:plevin@camtech.net.au)

**The Association For The Welfare of Child Health (AWCH) 8<sup>th</sup> National Conference.** 'Children on the Margin'. 11<sup>th</sup>-12<sup>th</sup> October 2001. Powerhouse Museum, Sydney, NSW. Contact (02) 9956-8333 or email [confact@conferenceaction.com.au](mailto:confact@conferenceaction.com.au)

**3<sup>rd</sup> National Conference on Spirituality & Disability.** 18<sup>th</sup>-21<sup>st</sup> October 2001 in Melbourne. Contact (03) 9251-5276 or email [andy.calder@vic.uca.org.au](mailto:andy.calder@vic.uca.org.au)

**Access Expo 2001.** 25<sup>th</sup> – 27<sup>th</sup> October, Royal Exhibition Building Melbourne.

Display of products & services for rehabilitation, disabled and aged care. Contact (03) 9415-1222 or [access.expo@paraquad.asn.au](mailto:access.expo@paraquad.asn.au)

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**Donor Conception Support Group Social Events** – 23<sup>rd</sup> September for NSW, contact (02) 9663-2565. 28<sup>th</sup> October for Victoria, contact (03) 9884-7457.

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**Neuromuscular Conference** – 7<sup>th</sup>-9<sup>th</sup> November 2001. Auckland, New Zealand. Contact: Muscular Dystrophy Association. PO Box 16-238 Sandringham, Auckland New Zealand. Phone/Fax: (09) 815-7260. Email: [Denise@mda.org.nz](mailto:Denise@mda.org.nz)

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AGSA

“Making the Right Connections  
since 1988”

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## 10 Top Reasons to Attend a Support Group

10. There are usually empty seats, refreshments are often served, and there is always a restroom nearby.
9. If you don't want to talk, you can just listen.
8. People discover it is okay to care about oneself.
7. Knowledge is power.
6. Anger, grief and loss are common feelings that are okay to talk about.
5. Hope and encouragement are important gifts of the group.
4. Shared feelings, not solutions, give the group power.
3. Living with difficult life situations colours life, but you choose the colour.
2. One of the rewards of life is that no one can seriously help another without also helping themselves.
- 1. Where else can you go where people will understand what you are talking about?***

(Blepharospasm Support Group  
Newsletter June, 2001)