

# ASSOCIATION OF GENETIC SUPPORT OF AUSTRALASIA NEWSLETTER

*Making the right connections since 1988*

DECEMBER 2013 ISSUE 101 ISSN1033-8624

Funded by **NSW HEALTH** & Ageing Disability and Home Care (ADHC)

*The AGSA newsletter is kindly sponsored by Genea*



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### MISSION STATEMENT

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

## EDITORIAL

Dear Members,

The Christmas bells are ringing and the shops and car parks are being blasted with Christmas carols as people rush around in the pre Christmas frenzy. Tempus fugit. Fortunately Christmas it is also a time when we stop, relax, enjoy the company of friends and go on holiday somewhere or just stay at home. AGSA will be closed from 20<sup>th</sup> December 2013 until 8<sup>th</sup> January 2014.

I would like to welcome to the AGSA committee, Kate Dunlop, Director for the Centre of Genetic Education and Anne Cutler Co-ordinator of Association of Welfare of Children in Hospital. Also I would like to give a big thank you to Richard Petrie, Brenda Phillis and Carolyn Shalhoub who have agreed to stand for another year. Also thank Michael Cori our retiring President for his wonderful support of AGSA over the last eight years.

Catherine Spinks our Research Officer will be finishing up on 20<sup>th</sup> December as her research is now completed. We have all enjoyed working with Catherine and she has done an excellent job on researching the "unmet needs of rare diseases in the Hunter community". We wish her well in her next ventures.

AGSA is looking for a Deputy Director to commence work in February 2014. The position will involve learning everything about how AGSA operates. If you are interested please give me a ring or email me and I will send you a job description.

On 8<sup>th</sup> September AGSA, in partnership with Prof Ingrid Winship, and Louisa di Pietro of GSNV held a Hereditary Hemorrhagic Telangiectasia (HHT) seminar at Royal Melbourne Hospital Melbourne. Approximately 60 people attended. The HHT Alliance website was launched so HHT people can now register on line.

<https://www.hht.org.au/Home>

On 13<sup>th</sup> September AGSA in partnership with GSNV held a Rural Outreach Seminar in Albury plus a sibling workshop. Dougie Herd from NDIS gave an excellent presentation and a Dad's personal story was very well received. Genetic counsellors from Melbourne also gave interesting presentations.

On 13<sup>th</sup> October, AGSA held its 12<sup>th</sup> BRCA Information Day which was once again a huge success with over 100 people attending of which 73% were newcomers. Thank you to the Genetic Counsellors and the doctors who gave up their time freely to plan, present and attend the day.

I would like to wish you all a Merry Christmas, a Happy New Year and a safe holiday season. Until 2014

DIANNE PETRIE OAM

## LETTER TO THE EDITOR

Dear Members:

I wanted to share with you some exciting news from the world of chromosome research from Dr. Iosif Lurie, CDOs consulting geneticist. Dr. Lurie provides CDO with the latest publications on research and development.

From Dr. Lurie:

As you know many new microdeletion syndromes are caused basically by deletion of one "main" gene. Deletions of several neighboring genes have no or very little clinical significance. One of such syndromes is Phelan-McDermid syndrome (del 22q13) where loss of SHANK3 gene is the main causative factor.

A new article by Shcheglovitov et al. which just appeared in "Nature" shows that increasing SHANK3 expression or treating cells by insulin-like growth factor can restore normal cellular function. Of course it was done in cell culture, and there is a long way before clinical application, but it is a potentially totally new path for treatment of patients. Usually I do not send you "non-clinical" articles, but this one is so interesting and so exciting that I feel necessary to share it with CDO.

The described article is highly technical but if you wish a copy please contact me directly at

[linda.sorg@chromodisorder.org](mailto:linda.sorg@chromodisorder.org)

Best wishes, Linda

Linda Sorg

President

Chromosome Disorder Outreach, Inc.

P.O. Box 724

Boca Raton FL 33429-0724



### GENETIC AWARENESS WEEK LAUNCH 18<sup>th</sup> September

Approximately 100 people attended to celebrate Associate Professor Kris Barlow-Stewart's contribution to genetic education and to hear Professor John Christodoulou's talk on whole genome sequencing. Below L-R Joanne Lind, Dianne Petrie A/P Kris Barlow-Stewart.



## 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.

**Apert syndrome – prenatal diagnosis**  
**Combine Immunodeficiency due to DOCK8 deficiency**  
**CDKL5 - <http://www.cdkl5.com>**  
**Foetal Valproate Syndrome**

### SAFDA Support After Foetal Diagnosis of Abnormality.

#### Contact Details:

Royal Hospital for Women Barker Street Randwick NSW 2031. Phone (02) 9382 6670.

SAFDA (Support After Fetal Diagnosis of Abnormality) has been established in South Australia to support parents and families before and after a termination of pregnancy following the diagnosis of an abnormality. The group is run by parents who have experienced this type of pregnancy loss supported by genetic counsellors.

Contact :

Anne Baxendale

Genetic Counsellor

South Australian Clinical Genetics Service

Women's and Children's Hospital

Phone: (08) 81617375

Email: [anne.baxendale@health.sa.gov.au](mailto:anne.baxendale@health.sa.gov.au)

### SAFDA VIC

At the time of diagnosis of a fetal abnormality, testing and decision making can seem overwhelming. There may be limited time to reflect on what has happened. The experience may be so profound that it takes months for its' meaning to sink in. Family and friends may offer lots of support at this critical time. However, as time passes and others move on with their lives, many parents find it helpful to talk with others who have had a similar experience, to share and reflect on what has happened.



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## NEWS IN RARE DISEASE AROUND THE WORLD

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### BREAKING GROUND AT SITE OF NEW HOUSING FOR PEOPLE WITH DISABILITY

Media Release  
8 August, 2013.

**Evolve Housing**, one of the State's largest community housing providers, in partnership with **RASAI** (Ryde Area Supported Accommodation for Intellectually Disabled) are pleased to announce that Federal Parliamentary Secretary for Housing and Homelessness, Senator Doug Cameron and NSW Minister for Ageing and Disability Services and Minister for the Illawarra, John Ajaka, will officially break ground today (12.30pm **Thursday August 8**) at the site of a new innovative housing complex at North Ryde, for people with disability.

Senator Cameron and Minister Ajaka will be joined by Minister for Citizenship and Communities, Minister for Aboriginal Affairs and the Member for Ryde Victor Domionello, as well as the City of Ryde Mayor and other Councillors.

The development, located in Smalls Road, was funded by the Federal Government, under the Supported Accommodation Innovation Fund (SAIF) and a grant from the NSW Government. Evolve Housing was awarded the high level Platinum Level compliance for the group home design which incorporates Universal Housing Design principles and supports the requirements of the Livable Housing guidelines.

Senator Cameron said: "This innovative development will provide better access to community-based accommodation and a supportive living environment for people living with disability.

"I'm delighted the Federal Government has provided a \$3.7 million grant for Evolve Housing to construct 15 new supported accommodation places in Ryde, which is in addition to our unprecedented investment in disability services through the new DisabilityCare."

Minister Ajaka said: "The NSW Government is committed to the provision of accommodation that supports people with disability to live as independently as possible.

"This enables people to plan for the future. For families and their sons and daughters, knowing that they are living in a place which is centred around their needs and aspirations, is incredibly important."

### FDA AWARDS 15 GRANTS TO STIMULATE DRUG, DEVICE DEVELOPMENT FOR RARE DISEASES

FDA NEWS RELEASE

Oct. 21, 2013

The U.S. Food and Drug Administration today announced it has awarded 15 grants totaling more than \$14 million to boost the development of products for patients with rare diseases.

The Orphan Drug Act was passed in 1983 to stimulate the development of products to treat rare diseases and conditions. For drugs, a disease or condition is considered rare if it affects less than 200,000 persons in the United States. For medical devices, a disease or condition is considered rare when it occurs so infrequently in the United States that there is no reasonable expectation that a medical device for such disease or condition will be developed without assistance. There are about 6,800 rare diseases and conditions, according to the National Institutes of Health. In total, nearly 30 million Americans suffer from at least one rare disease.

A panel of outside experts with experience in the disease-related fields reviewed applications for the grants, which will be administered through the FDA's Orphan Products Grants Program. The grant recipients are:

- Leonide Saad, Alkeus Pharmaceuticals, Inc., Phase 1 Study of ALK001 for the Treatment of Stargardt Disease—about \$167,000 for one year
- Parinda Mehta, Children's Hospital Medical Center Cincinnati, Phase 1 Study of Quercetin for the Treatment of Fanconi Anemia—\$600,000 over three years
- Diva De Leon, Children's Hospital of Philadelphia, Phase 2A Study of Exendin for the Treatment of Congenital Hyperinsulinism—about \$759,000 over three years
- Soma Jyonouchi, Children's Hospital of Philadelphia, Phase 1 Study of IL-2 for the Treatment of Wiskott-Aldrich Syndrome—\$600,000 over three years
- Dwight Koeberl, Duke University, Phase 1/2 Study of Clenbuterol for the Treatment of Pompe Disease—\$400,000 over two years
- Leslie Kean, Emory University, Phase 2 Study of Abatacept combined with Calcineurin Inhibition and Methotrexate for Prophylaxis of Graft versus Host Disease—about \$1.6 million over four years



- Todd Levine, PNA Center for Neurological Research, Phase 2B Study of Memantine for the Treatment of Amyotrophic Lateral Sclerosis—about \$990,000 over three years
- Joseph Reynolds, Spineform LLC, Phase 2 Study of the HemiBridge System for the Treatment of Idiopathic Scoliosis—\$1.6 million over four years
- Jonathan Davis, Tufts Medical Center, Phase 2 Study of rhCC10 to Prevent Neonatal Bronchopulmonary Dysplasia—about \$1.6 million over four years
- Glenn Furuta, University of Colorado Denver, Phase 2 Study of Esophageal String Test in Diagnosing Eosinophilic Esophagitis—about \$400,000 over two years
- Johan Van Hove, University of Colorado Denver, Phase 1/2 Study of Taurine for the Treatment of Cystathionine Beta-Synthase Deficient Homocystinuria—about \$400,000 over two years
- Laurence Cooper, University Of Texas MD Anderson Cancer Center, Phase 1 Study of Umbilical Cord Blood Derived CD19 Specific T cell Therapy in the Treatment of Advanced B Cell Malignancies—\$600,000 over three years
- Karl Anderson, University of Texas Medical Branch Galveston, Phase 2 Study of Hemin for the Treatment and Prevention of Porphyria Attacks—about \$1.5 million over four years
- Christopher Goss, University of Washington, Phase 2 Study of Intravenous Gallium Nitrate for the Treatment of Cystic Fibrosis—about \$1.6 million over four years
- Michael Debaun, Vanderbilt University, Phase 2 Study of Montelukast for the Treatment of Sickle Cell Anemia—about \$1.59 million over four years

“The FDA is committed to fostering and encouraging the development of products for rare diseases, most of which have no available or adequate treatments,” said Gayatri R. Rao, M.D., J.D., director of the FDA’s Office of Orphan Product Development. “The grants awarded this year support studies in very vulnerable, difficult-to-treat populations who have no available options.”

The FDA’s Orphan Products Grants Program was created by the Orphan Drug Act to promote the development of products for rare diseases. Since its inception, the program has given more than \$300 million to fund more than 530 new clinical studies developing treatments for rare diseases and has been used to bring 50 products to marketing approval.

### STATEMENT FROM NORD: FDA Orphan Products Grants Announced

The Food and Drug Administration yesterday announced the awarding of 15 grants with a total value of more than

\$14 million for development of drugs and devices for rare diseases, including several diseases covered by NORD member organizations.

The diseases for which the drugs and devices are being developed include congenital hyperinsulinism, Pompe disease, eosinophilic esophagitis, porphyria and other diseases affecting patients and families served by organizations that are members of NORD.

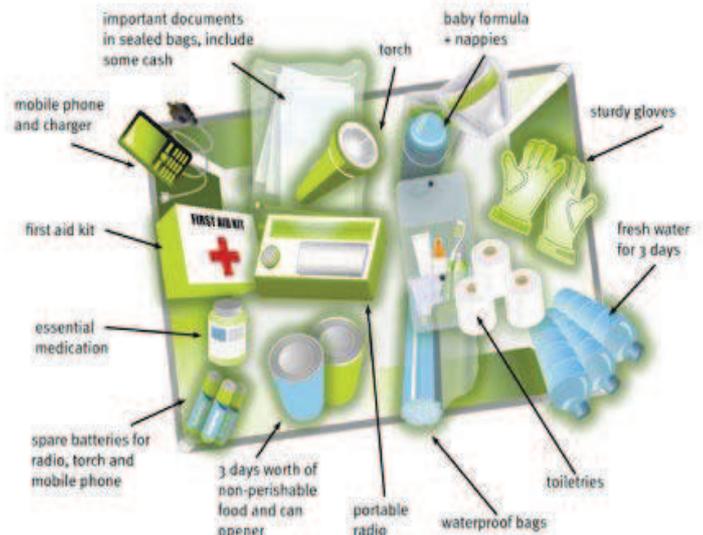
FDA’s Orphan Products Grants Program was established by the Orphan Drug Act to promote the development of products for rare diseases. The program has awarded grants worth more than \$300 million over the years for more than 530 clinical studies.

The Orphan Products Grants Program has provided funding that helped make possible the development of 50 products that ultimately received marketing approval for patients with rare diseases.

### 8 TIPS TO HELP PEOPLE WITH A DISABILITY PREPARE FOR STORM SEASON

Natalie Corbett - Friday, November 15, 2013  
RediAssist Blog

Severe weather events bring stress and uncertainty to everyone as we worry about the safety of ourselves, our loved ones, and our property. For people with disability and their carers, it’s even more important to consider the kinds of things that could go wrong in a storm, cyclone, flood or bushfire and prepare in advance.



Here are a few simple tips to help you and your loved ones prepare this storm season.

#### 1. Prepare an emergency kit

Medication and assistive devices should be top of



your list of what to pack, as well as prescriptions and other medical documentation. Read the [Get Ready Queensland Emergency Kit fact sheet](#) for what else to include.

## 2. Create an evacuation plan

Wheelchairs, medication and assistive devices are key to your safety and wellbeing, so make sure people helping you know to grab them. Don't wait until the last minute to evacuate - get out early while help is still available.

## 3. Arrange back-up power sources

Do you own electric mobility or medical equipment, or keep your medication in the fridge? Storms can cut power, so invest in a generator or find out if your equipment can run on batteries.

## 4. Organise transport

Can you get around if taxi and public transport services are disrupted? These services may be inaccessible for a number of days – possibly even months if networks are severely damaged.

## 5. Speak with carers

If your carer or support worker can't reach you, can you cope? Alternative care arrangements may need to be organised well before severe weather hits.

## 6. Get help to prepare your property

Trim overhanging trees, clear gutters and secure outdoor furniture well before a storm hits. Neighbours, family or service organisations can help with this and prevent damage to your property.

## 7. Be heard!

Some local councils or emergency services provide a registration service for residents with a disability and provide assistance for severe weather. During the 2011 Queensland floods, the [Queenslanders with Disability Network](#) held a 'phone around' to check in with contacts, ask your local support organisation if they'd consider doing the same.

## 8. Listen out

Pay attention to warnings and give yourself plenty of time to prepare. Sign up for [early warning alerts](#) to receive weather updates on your phone or email. Have a battery operated radio to stay informed throughout the emergency. For more information on disaster preparedness, visit [Get Ready Queensland](#) and complete the [Red Cross Emergency REDiPlan](#) for people with disability.



# PERSONAL STORY

## Travelling to the USA

### Seanne Lavender and the Lavender family

We arrived home last week after accomplishing the seemingly impossible.

We (Annalise, Gaby and myself) flew to Orlando USA to attend the inaugural international meeting of families who have children with Pyruvate dehydrogenase deficiency (PDH) at Walt Disney World, Florida.

The task was not easy. We planned everything as best as we could.

Gaby needed lots of sedation on the very full flights, but otherwise all went "well".

Meeting other families who all have children on a ketogenic diet, meant that her formula could be provided by the locals, (saving us 24 kg in extra luggage!)

It was absolutely wonderful to meet all these families (23 in all, several of whom had already lost their children). We shared many stories, lots of hugs and many tears.

We had 2 formal dinner meetings, both held in function centres within the Disney theme parks (complete with Disney character appearances each time!)

The second dinner was addressed by 3 well known /published USA Doctors who have done research into mitochondrial disorders and PDH

Dr Stacpoole who ran the DCA trial in Florida, Dr Douglas Kerr who has been passionate about PDH research (and is about to retire) and Dr De Brosse who is taking over from Dr Kerr and has a keen interest also in PDH.

There was much talk regarding development variations, outcomes, dietary management (all were on various types of ketogenic diets) and medication regimes. But above all, we developed a close network of families who share a similar path.

It has taken 17 years, but finally I can say, we have a network of PDH friends from over the globe now with whom we can share our highs and lows on a very intimate level.

Thanks to everyone who supported us in our desire to go (be it a crazy idea or not!!)

**PS** And for the record, we will not be taking Gabs on any more long international flights! Qantas were wonderful, but economy seating just isn't the place for anyone to be comfortable on a 14 hour flight, let alone Gaby! My back may never recover!

**Fiji will be far enough from now on I feel!**



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## CONFERENCES & EVENTS

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### **Williams syndrome conference**

**18-21<sup>st</sup> September 2014**

**Macquarie University**

**Contact AGSA for details.**

### **Genetic Awareness Week Launch**

**3<sup>rd</sup> September 2013**

### **BRCA Information Day**

**Sunday 12<sup>th</sup> October 2014**

**Sebel Surry Hills**

**Contact AGSA for details.**

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## USEFUL SERVICES

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### **Taxi Transport Subsidy Scheme**

The Taxi Transport Subsidy Scheme, also known as “the TTSS” or “the Scheme”, is administered by Transport for NSW.

The Scheme was introduced in 1981 to assist residents of New South Wales who are unable to use public transport because of a qualifying severe and permanent disability.

The Scheme subsidises the travel cost of approved applicants, allowing them to travel by taxi at half fare. The maximum subsidy that can be claimed is \$30.00 per trip. Participation in the NSW Scheme is not means tested however applicants must meet strict eligibility criteria.

Receipt of an aged, invalid, blind or other pension, or membership of a similar Scheme in another State or Territory, will not automatically qualify you for participation in the NSW Scheme.

In assessing your eligibility, we use an independent medical advisor to review your medical condition(s). We consider the recommendation of the medical advisor in conjunction with your overall application to decide whether or not you are accepted into the Scheme. Our privacy policy is also included on the application form. If your application is successful you will be required to comply with the Terms and Conditions applicable at the time you travel.

If approved to the Scheme you will receive a book of travel docketts. Taxi drivers will accept a docket from you in payment of 50% of the total fare up to a maximum of \$30 per trip. You must pay the balance of the fare, e.g. using cash or a credit facility.

The total fare charged by the taxi driver will be no greater than the fare charged to the general public.

There is no limit to how many trips you may undertake and you can use your docketts for all your taxi travel. You can go where and when you want (within NSW) such as travelling to and from work, the shops, social activities and leisure pursuits. There are also special vouchers to use if you are visiting another State or Territory.

Wheelchair accessible taxi (WAT) drivers receive a separate payment for transporting TTSS participants travelling in a wheelchair in a wheelchair accessible taxi. This payment is made by the Government and is not added to your fare.

### **To apply for admittance to the Scheme:**

1. Obtain a copy of the approved application form by phoning the toll free telephone number on the back cover of this booklet;
2. Complete PART A of the application form;
3. Take the application form to your doctor who will complete the remaining relevant questions on the form (PART B) and provide their details (PART C);
4. Send the completed application form to the mailing address on the form.

You and your doctor need to provide all required and requested information on your application form as specified. If your application form is missing information we will return it to you for correction and the assessment of your eligibility to participate in the Scheme will be delayed.

If you are assessed as being eligible for the Scheme you will be issued with a book of travel docketts containing your name and account number. You must present one docket to the taxi driver when you are claiming the taxi transport subsidy.

Information about your use of the Scheme including details of all journeys, will be collected and held by Transport for NSW and/or the payment processing provider or their contractors. This information will be used in the administration of the Scheme.

The information will also be used to identify abnormal travel patterns which may result in you being requested to provide further information about your travel and circumstances.



Should you require any further information about our privacy policy you should contact: **The Privacy Officer Transport for NSW PO Box K659 Haymarket NSW 1240 Tel: 02 8202 3768 E: gipa@transport.nsw.gov.au**

## MOTHERSAFE

MotherSafe is a free telephone service for the women of NSW, based at the Royal Hospital for Women, Randwick.



MotherSafe provides a comprehensive counselling service for women and their healthcare providers concerned about exposures during pregnancy and breastfeeding. Such exposures may include:

Prescription drugs  
Over-the-counter medications  
Street drugs  
Infections  
Radiation  
Occupational exposures

Some women may be offered face-to-face counselling appointments at the MotherSafe clinic which is held each week at the Royal Hospital for Women, Randwick. An interpreter can be arranged for women who do not speak English.

### **MotherSafe Contact Numbers**

Phone: 9382 6539 (Sydney Metropolitan Area)  
Phone: 1800 647 848 (Non-Metropolitan Area)  
Fax: (02)9382 6070

Interstate and overseas consumers should contact their local service.

### **Non-English Speakers**

Non-English speakers can access an interpreter through TIS National by phoning 131 450. You should tell the operator the language you speak, as well as the name and phone number of the organisation you wish to contact. There is no charge for interpreter costs when contacting a government-funded service or agency (such as MotherSafe or the Royal Hospital for Women).

### **Hours of Service**

**Monday–Friday 9am–5pm**  
(excluding public holidays).

# PROFILE

## A – Z OF GENETIC CONDITIONS

### ANGELMAN SYNDROME.... Did You Know???

Angelman Syndrome (AS) is a rare neuro-genetic disorder named after an English paediatrician, Dr Harry Angelman, who first described the syndrome in 1965. The AS gene UBE3A was identified in 1997. Rare - affects 1:20,000

#### Features always seen:

- Severe developmental delay e.g. delays in sitting and walking, delays in fine and gross motor skills development and delay in toilet training
- Profound speech impairment – no speech or minimal (receptive & non verbal communication > than verbal ones)
- Movement or balance disorder and ataxia of gait (sometimes stiff jerky limbs)
- Behavioural uniqueness; frequent laughter/smiling happy demeanour, easily excitable personality, often with hand flapping movements, short attention span and hyperactivity

#### Features usually seen, (80%)

- Small head size
- Seizures – onset often before 3 years
- Abnormal EEG (brain wave irregularity)

#### Features sometimes seen (20 – 80%)

- Protruding tongue
- Tongue thrust; suck /swallow disorders
- Feeding problems during infancy
- Wide mouth, widely spaced teeth
- Frequent drooling/excessive chewing & mouthing
- Sleep disturbance
- Scoliosis
- Strabismus( crossed eye)
- Fascination with water
- Wide based walking gait (feet turned out)
- Hypo pigmented skin, light hair & eye colour compared to family

#### Support

**Angelman Syndrome Association of Australia Inc.**  
PO Box 554



Sutherland NSW 1499  
Email: President @angelmansyndrome.org  
[AngelmanClinic@sesiahs.health.nsw.gov.au](mailto:AngelmanClinic@sesiahs.health.nsw.gov.au)

**Developmental assessment Service St George Hospital**  
Kogarah  
Phone: (02)8566 1222

**Is there a cure for Angelman Syndrome?**

Not at the moment, but researchers are working on it.  
See: [research](http://www.angelmansyndrome.org/research.html)  
<http://www.angelmansyndrome.org/research.html>

Some symptoms can be treated. The condition is permanent but is not degenerative. AS children can look forward to a normal lifespan. have an irregular chromosome 15. Recent studies have identified an AS gene on chromosome 15. Research is continuing worldwide on the complex genetics of AS to better understand why it occurs. Males and females are affected equally

When children with Angelman Syndrome are observed and studied, many educational and behavioural interventions have been shown to be effective in the areas of communication, behaviour modification, sleep disturbance, general conduct and social skilling.

**Physical and occupational therapies, speech and language intervention assist AS children.**



**AGSA would like to wish all it's members, families, individuals and readers a very Merry Christmas and a Happy New Year!!**

**AGSA has some great things planned for next year so we look forward to seeing you all then!!**



## AGSA NEWSLETTER ISSUES

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October 1995	20	Velo-Cardio-Facial syndrome
August 1995	19	Haemochromatosis/ Grief seminar
June 1995	18	Psychiatry & Genetics
March 1995	17	Sotos syndrome
February 1995	16	Gaucher disease/Lysosomal Storage Disorders
December 1994	15	Rare Chromosomal disorders
October 1994	14	Klinefelter syndrome
August 1994	13	Leigh disease
May 1994	12	Angelman syndrome
January 1994	11	Fabry disease
October 1993	10	Huntington's disease
Winter 1992	9	Ectodermal Dysplasia, Sanfilippo disease
March 1992	8	Marfan syndrome, Rett syndrome
March 1991	7	Tuberous Sclerosis, Noonan syndrome
Sept-Dec 1990	5 & 6	Osteogenesis Imperfecta, Ehlers-Danlos syndrome
June 1990	4	Turner syndrome, Achondroplasia
March 1990	3	Congenital Adrenal Hyperplasia, Biochem Genetics

November 1989	2	Antenatal diagnosis, Victorian Genetic Services
April 1989	1	Drawing a family health tree, NSW Genetic Services

## MILESTONES

AGSA first newsletter was published in 1989 with the help of the AGSA Committee and Margot Latham at the Children's Hospital Westmead.

In 24 years we have profiled the same genetic condition only twice each time with different personal stories.

Today AGSA's principle is still the same, peer support, information and contact with others and it is still what people want. This support gives relief from isolation and provides hope. AGSA has been providing this type of support for over 24 years.



**Under Jaguar Sun by Antonio Nava Tirado at the Broken Hill Sculpture Site NSW.** This sculpture depicts Duality; day and night where

night is represented by the Star of Venus, the mouth. Day is represented by the circle created by the Sun. **Background:** A successful sculpture symposium was held on this majestic hilltop (within the centre of the reserve) in 1993 by artists from around the world, under the direction of organiser and artist Lawrence Beck. 12 sandstone sculptures highlight the skyline, all with a story to tell. The Sculpture Site can be accessed via a 1 km walking trail starting from the Flora and Fauna Sanctuary car park in Broken Hill.

### Why this picture?

This symbolises the complexity and continuous role of caring and the beauty of unconditional love and the many personal stories. The sculpture also depicts a C for century – a hundred newsletters.

