THE•ASSOCIATION OF•GENETIC•SUPPORT OF•AUSTRALASIA

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MISSION STATEMENT

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

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EDITORIAL

I am extremely honoured to be elected President by you. Thank you for your support and confidence in me. From the old committee, Ros and Margot in particular, made a fantastic contribution, both being upstanding dedicated people who have really put AGSA on the map. I am sorry to see both of them also Sue and Judy going. The new committee has only had one meeting, but I can see the new blood will bring fresh life into our workings and I hope we can achieve a lot this year. Since we did not get extended funding from the Department of Health, we would like to get into some fundraising this year to make up for it. Dianne desperately needs a fax, and a few more hours a week could make a world of difference to what Dianne can achieve, another desk in the room would be convenient...Oh! The wish list goes on and on. Our Awareness Day this year is on 24th July, but we have many things planned both before and after this. I am looking forward to your support.

Best wishes,

DEBBIE REDELMAN

PEER SUPPORT/INFORMATION OFFICER'S REPORT

Our last AGM saw the re-election of the committee. A fond farewell was given for Ros Smith who was President for three years and Margot Latham, Treasurer, Sue Clough, Judy Rands, our Regional Representative and Sarah Bridge.

Ros has put her stamp on AGSA right from the beginnings in 1988 and through her leadership and strength AGSA is where it is today, an essential part of the community. Margot initially began as a committee member, rose to President

and latterly served as Treasurer for two years. All outgoing members have contributed enormously to AGSA and we will miss them.

I would like to take this opportunity of welcoming in the three new committee members. Sue Manass, Genetic Counsellor, Sydney Children's Hospital, David Lewis, Genzyme, Louise Scott, Regional Representative and Marlene Brightwell, Co-ordinator Rare Chromosome Disorders Support Group.

Last month I attended the annual Genetic Counsellor's two day workshop in Melbourne held at the Royal Women's Hospital. Day one of the workshop covered important issues such as "Cultural Aspects of Bereavement", "Children's Grief", "Adolescent and Family Issues of Grief" and "Ultrasound Diagnosis and Genetic Counselling" and "Pre-natal Grief". Day two involved case discussion group work, "The Neurology You Need to Know" and "Caring for the Care Givers". The talks over the two days were excellent and the case discussions gave me greater insights into my work. It was wonderful to put faces to voices heard over the telephone and to meet first hand genetic counsellors from around Australia.

On Saturday I attended the Neuronal Intestinal Dysplasia (NIDKIDS) Support Group meeting at the Murdoch Institute. NID is when nerve fibres in the outer bowel are deficient in substance P (SP) causing chronic constipation. Prof. John Hutson who is currently doing research into NID opened his labs to show the group around and to discuss his research. Helen Bird established the NIDKIDS Support Group in 1995 and Prof. Hutson and Helen will be giving talks at the various hospitals around the state on NID. They are expecting the group to grow very quickly as there are many undiagnosed cases.

I would like to extend my thanks to the Genetic team in Melbourne for a great two days and also to Helen and John for the opportunity of learning about their group and meeting the members first hand.

Very soon I will be travelling to Albury to the IDEAS Expo where AGSA will have a static display. On the Saturday morning I am holding a workshop on "The Effect of a Disability on the Family". I am looking forward to this and to meeting up with country families.

VALE

On a sadder note I am sorry to report Dr Tony Lipson, Paediatrician, Department of Clinical Genetics, New Children's Hospital, died suddenly during Easter. Tony was a man who stood behind many support groups and assisted AGSA in an advisory capacity over the last two years. I was assisted by Tony in the eighties when I established the Williams Syndrome support group.

On behalf of AGSA and our members we extend our sympathy to his family and colleagues. He will be greatly missed by many.

Best wishes,

DIANNE PETRIE

MESSAGE FROM THE DEPARTING PRESIDENT

Over the many years with AGSA I am particularly grateful for the partnership which has developed between the committee and the professionals throughout Australia and their belief in AGSA. Their support has played a big part in raising AGSA's profile. I would like to thank Margot Latham for her outstanding support as Treasurer and friend, Dianne Petrie for her commitment and the tireless energy she has applied to the position, the committee and a special thank you to our growing membership both here and overseas.

I have enjoyed my time with AGSA and I will certainly miss the day to day contact with families. I wish everyone the best for the future and particularly the incoming AGSA committee.

ROS SMITH

FROM THE DEPARTING TREASURER

I would like to take the opportunity to thank all the committee members, both past and present, for their support during my time as, firstly a committee member and then Treasurer of AGSA. I would especially like to thank the membership, who have shown through their continued association, that they see AGSA as a beneficial

and helpful organisation. I know that without the support of the membership the committee's task would be a very difficult one. Thank you to Dianne who has taken on the role of the first Peer Support and Information Officer with enthusiasm and vigour. To my dear friend Ros Smith, who like me has retired from the committee, a special thank you for the hours spent together and the laughter that so often ensued. Having moved to Melbourne I am certainly missing the many friends that my time with AGSA has given me the pleasure to meet. I hope to keep in touch and visit when possible and will certainly stay a loyal member of AGSA. So once again thank you to the many people whom I have had the pleasure to meet and work with.

MARGOT LATHAM

PROFILES

DEBBIE REDELMAN, JP, - President

Debbie has been a professional on our committee for three years, with a Science degree in Human Genetics and Psychology. She has worked as a research assistant reporting on birth abnormalities, a telephone counsellor, state co-ordinator of the Tourette Syndrome Association and is currently working on the Tay Sachs screening program. She is also a member of the Teratology Society and the Human Genetics Society. Debbie is married with three children aged 8, 6 and 4. Her other interests include cooking, craft, the stockmarket, handwriting analysis, theatre and walking.

RICHARD PETRIE - Vice President

Richard has been on the AGSA committee for about eight years and believes the organisation has a unique role in meeting the needs of a significant segment of the community.

Richard has a daughter with Williams syndrome, now aged thirteen and at the time of diagnosis doctors knew of only two other families in Australia with a WS child.

There was little information available on the condition and very poor information on what support services were available. Everything seemed very difficult. With his wife, Dianne, they set up the Williams Syndrome Support Group in 1985 which now has over 160 members nationally. Richard believes if AGSA had been in existence many aspects of living with a child with a disability could have been so much easier.

Richard has a background in pharmaceutical sales and marketing and works as a marketing contractor.

DAVID LEWIS - Pen portrait of a Wanderer

- Committee Member

David Christopher Lewis, was born on Christmas Day too many years ago to remember. Married, to Marie. They have two daughters Erica and Elizabeth and a son Christopher.

David has a rare genetic disease inherited from his mother named closed angle glaucoma and is exiled by choice from England and by circumstances from Queensland!

David spent most of his life in the export side of the Pharmaceutical Industry, during which time David was lucky enough to visit and enjoy over 80 countries and their people.

David currently works for Genzyme Corporation who manufacture and sell Ceredase and Cerezyme for the treatment of a rare autosomal recessive disease Gaucher's disease. One of the lysosomal storage diseases more common but less well known than Tay Sachs or Niemann-Pick's. He is responsible for the company's business in Australia and New Zealand. A major hurdle is funding.

David feels very committed to the patients and is convinced that rare diseases, especially with expensive treatments need strong umbrella group lobbies like AGSA and the proposed Lysosomal Storage Diseases Group if they are to get funding. David asks why is the government subsidising unnecessary antibiotic use and elective oral contraceptives which most of the population could afford for themselves but not life saving therapies that individuals can't afford to pay for?

MARLENE BRIGHTWELL - Committee Member

Marlene is a qualified piano teacher. Marlene is interested in Music Therapy and has worked using this skill with the playgroup at the Shepherd Centre, Sydney University.

Marlene's daughter was born with 18q- syndrome. The desire to meet other children with the same condition, meant travelling to Chicago in 1994, for the first Annual Chromosome 18 Registry & Research Conference. Realising the benefits of meeting other families, she decided to help with the formation of the Rare Chromosome Disorder Support Group of Australasia. Marlene is presently co-ordinating the group. Marlene also helps out in the AGSA office when needed.

BRENDA PHILLIS - Committee Member

Mother of three daughters 23, 20 and 17 all affected by Ehlers Danlos syndrome. Brenda has been the secretary of AGSA since its original working in 1987, except for a two year break. Brenda became involved because she felt the real need for an organisation such as AGSA after finding it difficult, upon diagnosis, to obtain information, contact etcetera as there was no support group.

SUE MANASS- Committee Member

Susan Manass is attached to the Medical Genetics Unit at the Prince of Wales Children's Hospital in the capacity of Genetic Counsellor. She has been employed at POWCH (now Sydney Children's Hospital - Randwick) for three years as a counsellor, and previously for 14 years in the Cytogenetic Laboratory as a scientist. Needless to say Sue's special interest is in chromosomes and chromosome abnormalities.

JAN CAMERON SMITH - Committee Member

Jan is a qualified laboratory technician (1970); and gained an Arts Degree (English & Linguistics in 1986, completed a post-graduate diploma in Arts (Library and Information Science) - librarianship in 1992.

Jan has Osteogenesis Imperfecta (O.I.-Brittle Bones) and is presently employed full-time as the sole librarian in a corporate organisation. From 1984-1995 Jan was elected to the Board of the Osteogenesis Imperfecta Society of Australia.

Jan has been involved with AGSA as a Committee Member from its pre-planning stages, to date, with a gap from 1990-1992.

Jan is a member of the Uniting Church -Synod Ministry to People with Disabilities team and she belongs to People with Disabilities Inc. This gives her more access to adults and a bigger forum with which to address the social issues facing adults with chronic degenerative conditions.

LOUISE SCOTT - Non -metropolitan Representative

Louise and her husband Stuart have one daughter now Martine. Martine's sisters, Jacqueline and April were affected by Spinal Muscular Atrophy Type I - Werdnig-Hoffman Disease . Louise has been a member of AGSA for the last six years and she is a member of SAFTA (Support After Foetal Diagnosis of Abnormality) Support Group. If anyone wishes to contact Louise just to have a talk please ring 069 202260 after hours or leave a message. Louise looks forward to meeting up with the rural families.

NB Mona Saleh will be profiled in the next newsletter.

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LETTERS TO THE EDITOR

The following is a summarised letter from a person who sought AGSA's assistance.

8th March 1996

Dear Dianne,

I spoke to you in late December and early January about our son recently diagnosed with Klinefelter.

When I spoke to you, Dianne, things looked very dark indeed to me. There wasn't much light coming into that long tunnel. The information you provided and the people you gave me to contact - particularly the Social Worker - were very helpful indeed. Our outlook is very rosy...especially by comparison with last year's dramas. I have had several long contacts by telephone with the Social Worker - she is informative and supportive without being intrusive.

Thank you, Dianne. You and your organisation are wonderful sources of information and networking.

Yours sincerely,

S.P.

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18th April l996

Dear Members,,

The CMT (Neurological disorder affecting arms & legs) Association would like to share with another support group available office space at Concord Hospital. If you are interested please contact :- Kay Boreham, President,

Charcot- Marie Tooth Association on 587 3150

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RESEARCH

DISCOVERY OF FRIEDREICH'S ATAXIA MUTATION

A new trinucleotide repeat disease

An Italian research group (Campuzano et al) has found the mutation causing Friedreich's Ataxia (Science 8 March). Most cases are due to an expanded GAA repeat: This is the first GAA repeat causing a disease. A few percent of cases have point mutations on one chromosome, combined with the expanded repeat on the other. The diagnostic test is being established at the Murdoch Institute, Melbourne and at the Molecular Medicine Laboratory, Concord Hospital, Sydney. This breakthrough means that definite diagnosis and carrier testing will be available at last.

This information was kindly supplied by A/Prof. Garth A Nicholson, University of Sydney, Department of Medicine, Molecular Genetics & Medicine Laboratory.

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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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GARDENER SYNDROME

A family would like contact for their son who has this condition. Contact AGSA for details.

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JUVENILE PAGET DISEASE (CONGENITAL HYPERPHOSPHATASIA)

AGSA has received a letter from an American family seeking contact with another family for their daughter. Contact AGSA for details.

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IMMOTILE CILIA SYNDROME (CILIARY DYSKINESIA, KARTAGENER SYNDROME)

There is a British Support Group for this condition however AGSA has received a request for contact with a family in Australia. If you know of anyone please contact the AGSA office for details.

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PAROXYSMAL DYSTONIC CHOREOATHETOSIS

A gentleman would like contact with another person who has this condition. Please contact AGSA for details.

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PEHO SYNDROME

A mother would like contact for her daughter with this condition. Please contact the AGSA office for details.

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POLOSTOTIC FIBROUS DYSPLASIA

A family for like contact for support and information. Please contact the AGSA office for details.

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RESOURCES

REHAB 4 KIDS - Paediatric Positioning & Mobility Products

Kid-Karts have a Sydney Showroom. Rehab 4 Kids have the sole distributor arrangements in place to import from Kid-Kart Montana, USA. The Karts are light, the seating section is removable in most cases and in all other situations the unit folds down for transportation. For further information contact: Judie Stephens.130 Letitia Street, Oatley NSW 2223, Tel: Mobile 0419 393 393

CONFERENCES

AUSTRALIAN CHEMICAL TRAUMA ALLIANCE

THEME: "REPRODUCTIVE EFFECTS AND GENETIC RESULTS OF EXPOSURE TO TOXINS"

A conference dealing with the hazards to children and to the unborn because of direct or parental chemical exposure.

Where: Coffs Harbour **When**: August 9, 10 & 11, 1996 **Speakers**: There will be a cross section of specialist from the fields of paediatrics, genetics, immunology and toxicology.

Fees: Public $85.00, ACTA Members $35

For further information or a registration form, please contact : ACTA Secretary, "Mimosa", Nullamanna 2360 Phone (067) 25 5521

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4th National Conference of the Association for the Welfare of Child Health (AWCH)

"Who cares?" Before and After The Hospitalisation of Children

10 -11 October 1996 - University of Western Sydney, Nepean, Westmead NSW

Call for Papers from parents and professionals and invitation to attend.

Theme - Wellness Rather Than Illness Model

Abstracts of 300 words required by May 6th 1996

For further information contact: Ms Lynn Shaw, National Development Officer, AWCH National Office, P.O. Box 113, Westmead NSW 2145 Ph: 633 1988 Fax: 636 1180

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.

However for this issue we provide here a list of genetic conditions represented by AGSA. This list represents the families, friends, parents, professionals and even grandparents who have contacted AGSA seeking support, contact and information.....

Aarskog syndrome
Acid Maltas Deficiency
Alagille syndrome
Albinism
Alkaptonuria
Alstroms syndrome
Androgen Insensitivity
Angelman syndrome
*Anorexia
Aperts syndrome
Aspergers syndrome

Batten's Disease
Beckwith-Wiedemann syndrome
Behr syndrome
BEPS

Canavans Disease
Carpal-tarsal osteolysis
Central Core Disease
CHARGE Association
Cerebellum Hypoplasia
Chondrodysplasia
Chromsome disorders
Cleido Cranial Dysostosis
Cohen syndrome
Congenital Adrenal Hypoplasia
Congenital Anodontia
Congenital Cone Dystrophy
Congenital dislocated hip
Congenital Myotonia
Congenital Prune Belly syndrome
Combined Immune Deficiency Disease
Cooleys Anaemia - Thalassaemia
Cornelia-de Lange syndrome
Cytochrome C. Oxidase Deficiency
*Creutzfeldt-Jakob Disease
Cystic Fibrosis
Cystinuria

*Dercum Colorosa

Ehlers-Danlos syndrome
Erythropoietic Protoporphyria
Ernster-Luft syndrome

Fabrys Disease
Facially disfigured
Facial Haemangioma
47, XXYY
48, XXYY
Fibromyalgia Hermaphroditism
*Fibrous Dysplasia
Fragile X syndrome
Fraser syndrome

*Gastric reflux
Gaucher Disease
Goldenhar syndrome
Haemochromatosis
Hereditary Fructose Intolerance
Homocystinuria
Huntington Disease
Hydrocephalus
Hydranencephaly
Hyper IGE syndrome
Hypoactive thyroid
Hypopituitism
Hypertrophic Cardiomyopathy
Hypoadrenal Hyperthroidism
Hypoplastic Primary Vitreous
Incontinentia Pigmentia
Intellectual disability
Immune deficiency disorders

Kabuki Make-up syndrome

Kawasaki syndrome
Klinefelter syndrome (47,XXY)

Landau-Cleffner syndrome
Larsen syndrome
Laurence-Moon-Biedl
Lissencephaly
Lymphas genphasia
Lymphodema
Lysosomal Storage Disorders

Marfan syndrome
Maple Syrup Urine Disease
MELAS syndrome
Microcephaly
Myotubular Myopathy

Neurofibromatosis
Noonan syndrome
Nonketotic Hyperglycaemia

Oculo-dento-digital syndrome
Opitz trigonocephaly
Osteopetrosis

Pallister-Hall syndrome
Paroxysmal Nocturnal Haemoglobin (PNH)
Pelizaeus-Merzbacher Disease
Peripheral Neuropathy (CMT Type II)
Phenylketonuria (PKU)
Picks Disease
Poland syndrome
Porphyria
Postnatal Psychosis
Potter syndrome
Pre-eclampsia
Progeria
Primary Immune Deficiency
Pseudoxanthoma Elastic (PE)

Reinfenstein syndrome
Rendu Osler Weber syndrome
Schwachmans syndrome
Severe Immune Deficiency
Sotos syndrome
Spina Bifida
Sprintzen syndrome (Velo Facial Cardio syn)
*SSPE
Stargardts Disease
Stickler syndrome
Sturge Weber
TAR syndrome
Tourette syndrome
Treacher-Collins
Trisomy 13, Trisomy 18
Triple X syndrome (47,XXX)
Turner syndrome (45,X)
Weaver syndrome
Weill-Marchesani syndrome
Werdnig-Hoffman syndrome
Williams syndrome
Wolf-Hirschhorn syndrome
VATER Association
Von Hippel-Lindau syndrome
** denotes non genetic conditions*

OVERSEAS CONFERENCES

Fanconi Anaemia Annual Family Meeting - May 16 - 21, Camp Sunshine, Point Sebago, Maine. U.S.A. Call for info: 1/800/838-4891

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Cornelia de Lange Syndrome Foundation Intern'l Conference - June 27-30 Sheraton Music City, Nashville, TN. Contact the Foundation, 60 Dyer Ave., Collinsville, CT . U.S.A.06022 Tel: 860-693-0159.

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Communicating Bad News, a scientific session for health professionals sponsored by the Support Organization for Trisomy 18,13 & Related Disorders (SOFT) - July 11; University of North Carolina at Chapel Hill. Contact Cynthia Powell, UNC Div. of Genetics, Campus Box #7250 Chapel Hill, NC 27599-250 U.S.A.

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STOP PRESS
CLEFTPALS NSW/NATIONAL AGM AND PROFESSIONAL PANEL MEETING
Date: 20th July 1996 Time: 9.15 am - 12.30 am
Venue: Lorimer Dods Theatre, Level 2, The New Children's Hospital, Hawkesbury Rd, Westmead.
Come along and hear a range of topics to be discussed by a panel of professionals from the New Children's Hospital Cleft Palate Clinic.
For further info., contact Roslyn Currey (02) 838 9313 or Helen Walker (02) 671 4753

AGSA'S CALENDAR OF EVENTS

You are invited to the following Seminars held by AGSA.

FRAGILE X SEMINAR

Date: 26th May 1996
Venue: 66 Albion Street, Surry Hills
Time: 10.00 am - 3.00 pm
SPEAKER: Hazel Robinson
Nurse Counsellor
Topic: "Female Adolescents and the Implications of being a carrier"
Lunch will be provided.
R.S.V.P. 15th May 1996

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SEMINAR ON SUPPORT GROUPS "THE PRACTICALITIES OF RUNNING A SUPPORT GROUP"

Speakers: Support group leaders and members Glenn and Geoff Fisher will be assisting with discussions.
Date: 2nd June 1996
Venue: 66 Albion Street, Surry Hills
Time: 10.00 am - 2pm
Registration Fee: $6.00
Lunch will be provided
R.S.V.P. 20th May 1996.

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SEMINAR ON ADULT CHRONIC DISORDERS

ISSUES: Discrimination in the workplace, transport, social issues, rehabilitation, transition phase between child and adult.
Date: 21st July 1996
Venue: 66 Albion Street, Surry Hills
Time: 11.00am - 4.00 pm
Lunch will be provided.
R.S.V.P. 2nd June 1996

GENETIC AWARENESS DAY 24TH JULY 1996

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FUNDRAISING EVENT AUGUST (date to be advised) AT THE HOLSWORTH GALLERY WOOLLAHRA

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Cut out and return to AGSA

Please circle and return to Dianne Petrie AGSA 66 Albion Street Surry Hills NSW 2010 or phone 211 1462 by the appropriate R.S.V.P. date.

Yes, I would like to attend

(1) Fragile X,

(2) Support Group Practicalities

(3) Adult Chronic Disorders Seminar

Name.....
.....
Address.....
Phone.....
Condition.....
.....

Yes, I would like to talk for 5 - 10 minutes at the Adult Chronic Disorders seminar on my experiences and abilities to overcome a certain situation.....
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.....
.....
.....

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

66 Albion Street
SURRY HILLS
NSW 2010

Tel: (02) 211 1462

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Support and Information Officer -
Dianne Petrie
Office Hours: 10.00am - 2.00pm
Monday - Friday

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President
Debbie Redelman
may be contacted on:
Tel: (02) 327 1048

Regional Contact
Louise Scott
"Dargle"
2 Norman Street,
The Rock NSW 2655
Tel: (069) 202260 (After Hours)

ANNUAL SUBSCRIPTION

Individual	$15.00
Group/Organisation	$30.00

Subscription Year
lst 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

* lobby government bodies, both Federal and State, for appropriate funding for genetic services