

THE ASSOCIATION OF GENETIC SUPPORT OF AUSTRALASIA INC.

FUNDED BY THE NSW HEALTH DEPARTMENT

NEWSLETTER

AUGUST 1999 ISSUE 43

MISSION STATEMENT

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

Reg Charity CFN15481 Tax Ref AF1595C/SF8566

Contents

Contact Corner: Blooms syndrome, Costello syndrome, Luvan syndrome, Holoprosencephaly, Hydranencephaly, Succinic Semialdehyde Dehydrogenase Deficiency, Pitt-Hopkins syndrome.

Profile: Support Groups
by Caroline Bowditch and Margaret Sahhar

**Come to AGSA's Fundraising Dinner
on 11th November 1999**

at Pepe's Restaurant

3 Beattie Street

Balmain

Time 7.00 pm

Cost: \$30.00 per head

BYO

**Make up a table by asking ten of your
friends and have a fun night.**

*AGSA would like to thank you for your
subscription renewals. Please note your receipt
should be enclosed with this newsletter.*

*A Special Thank You to those of you who gave
generous donations. It is greatly appreciated.*



*A special thank you to Dr John Collins
who regularly gives AGSA a generous donation
and recently some excellent text books. John
also attends many of the AGSA functions and
has been a great supporter over the years.
Thank you John.*

**AGSA's AGM Sunday 17th October 1999 at
10.00 am at the AGSA office.**

**Guest Speaker to be advised. Hope to see you
there.**

PEER SUPPORT /INFORMATION OFFICER'S REPORT

I tried cloning myself this month but was unsuccessful. A pity I could have used the other three people.

In July, I attended the launch of the Genetic Support Network of Victoria at the Murdoch Institute. To my surprise the audience was made up of many families I have been speaking to regularly over the years. It was wonderful to put faces to names and to meet their children for the first time. I look forward to working alongside Caroline Bowditch and Margaret Sahhar who have both been very supportive of AGSA. I know the Victorian families will benefit greatly from the formation of this organisation.

To all clinicians thank you for referring families to AGSA and I encourage you to hand AGSA's pamphlet to each new family. By doing so you provide more than just a diagnosis. AGSA provides direction and a very supportive network of resources and information for these families. The comment I often hear is "why didn't I know about AGSA earlier." If you need more pamphlets please give me a ring.

Recently, I attended the Human Genetics Society of Australasia Scientific Conference "Genetics into the New Millennium" at the Australian Technology Park, Redfern. As usual I found it very stimulating and an excellent time for catching up with many friends in genetic services and sharing information. I particularly enjoyed the Ethics debate on cloning, Huntington disease and predictive genetic testing in children and discrimination and eugenics in prenatal diagnosis.

With the AGM coming up we would like to hear from you if you wish to join our small but very productive fun committee.
Until then best wishes

Dianne Petrie



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.

BLOOMS SYNDROME

A family with a two year old boy would like to speak with other parents. Contact AGSA for details.

CONGENITAL ADRENAL HYPERPLASIA AND DUCHENNE DYSTROPHY

The family would like contact with others with either condition.

COSTELLO SYNDROME

A mother of a 23 year old daughter with this condition is keen to have contact with other families with the view of sharing information on development.

LUVAN SYNDROME

A family with two sons with this condition are eager to obtain more information and have contact with another family.

HOLOPROSENCEPHALY

A mother of a child with this brain abnormality is endeavouring to make contact in Australia with parents who may have had a child or pregnancy diagnosed with this disorder. Families will be meeting to obtain information and to participate in current research late September.

HYDRANENCEPHALY

A Queensland family who has a child with this condition is seeking communication with another family. Contact AGSA for details.

IVERMARK SYNDROME OR ASPLENIC SYNDROME

A mother of a one year old boy would like contact with others.

REFLEX SYMPATHETIC DYSTROPHY

AGSA knows of two families who would like any information on treatment and contact with others.

SUCCINIC SEMIALDERHYDE DEHYDROGENASE DEFICIENCY

A NSW family with a four and half year old boy with this condition would like contact with another family. The family already know of two other families in Australia. Contact AGSA for details.

PITT-HOPKINS SYNDROME

from Contact A Family, UK

A UK family has a 10 year old son with this condition and the Geneticist involved has given this as an "observational diagnosis" and wants to write up his case for publication. The boy concerned has learning disabilities, no speech, small stature, poor muscle tone, broad lips and respiratory problems (hyperventilates). Do you have any information on this syndrome or do you have any families who are seeking contacts? Contact AGSA for details.

1.

GROSVENOR EARLY EDUCATION CENTRE INC.

For young children with special needs

An early childhood intervention services for children aged 0 - 6 years and their families

For more information contact:

Director: Jan Patuiny

56 Liverpool Rd, Summer Hill NSW 2130

Ph/Fax: (02) 9799 6294

Email: grosvenor50@hotmail.com.

Homepage:

<http://www.geocities.com/Athens/Agora/8772/index.html>

Funded by: Department of Community Services; Aging and Disability Department, Commonwealth Intervention Support Program

PROFILE - A - Z

THE ROLE OF GENETIC SUPPORT GROUPS - WHEN THE FAMILY NEEDS HELP

There are several hundred patient and family advocacy and support groups. These range from very large and professional organisations which play a part in forming community attitudes and in research (such as the Anti-Cancer Councils) through to small groups which cater for the needs of a few isolated individuals or families affected by a rare disorder. As a clinician, how can support groups assist you?

Whether you are a GP or a specialist, you are an expert in your field. Your patients and their families will look to you for all kinds of advice. However, even if you have a good picture of the medical aspects of a condition, you are unlikely to know what it is like to live with it every minute. This is a particular problem for those individuals and families affected by a genetic disease, which is lifelong and often difficult to treat. Often, first hand experience is needed. Support groups can provide the "inside information", as they are made up of people who can frequently be considered to be the 'experts'

in their field - what it is like to have a specific genetic condition.

Support Groups in History.

Support groups have been in existence in our society for hundreds of years. They may pose as Mother's Clubs or male bonding groups, and may vary in their focus, but the underlying purpose is to provide mutual support to people with experiences similar to their own.

How can support groups assist your patients?

For those with a genetic disease, or have just had a child with such a disorder, the diagnosis is often dramatic and catastrophic. Practical resources are often needed, as well as information, and time to digest it. Support groups can pinpoint the types of sources that are available that could be most valuable to you and our patients, and can empower your patients and their families. They can help families to overcome the inevitable feelings of isolation and loneliness. Many families may feel they are the only ones in the world who feel the way they do and face this problem, but chances are they aren't. Support groups can offer people an opportunity to share their experiences, joys, frustration's and challenges. This support often occurs after hours in a comfortable and conducive environment. The groups can play an essential role in "normalising" a new diagnosis for a family. Perhaps most important, they provide an invaluable opportunity to network, and provide the knowledge and contacts to allow a family to avoid the 'reinventing of the wheel' which those with a child with a disability often find themselves involved in.

For young people affected by a specific condition, support groups can provide role models and can specifically focus on issues that may be age and/or condition specific.

Breaking Bad News

With any new serious diagnosis, such as that of a genetic disorder, comes the inevitable task of breaking of the bad news. Although support groups cannot take this process away from the clinician, they can assist a family through their grieving process. The following piece of writing

is frequently passed on to parents of children with newly diagnosed conditions - perhaps it could be of some use to you and your patients.

Welcome to Holland

When you are going to have a baby, it's like planning a fabulous trip to Italy. You buy a bunch of guide books and make wonderful plans. The Colosseum, Michaelangelo's David, the gondolas in Venice. You may even learn phrases in Italian. It's all very exciting!

After months of eager anticipation, the day finally arrives. You pack your bags and off you go. Several hours later the plane lands. The flight attendant comes and says, "Welcome to Holland"

"Holland?" you say. "What do you mean Holland? I signed up for Italy. All my life I dreamed of going to Italy"

But there's been a change of flight plan. They've landed in Holland and there you must stay. The important thing is that they haven't taken you to a horrible disgusting, filthy place full of pestilence, famine and disease. It's just a different place.

So you must go out and buy new books. And you must learn a whole new language. And you will meet a whole new group of people you would never have met.

It's just a different place. It's slower-paced than Italy. Less flashy than Italy. But after you've been there for a while and you catch your breath, you look around and notice that Holland has windmills. Holland has tulips. Holland even has Rembrandts. But everyone you know is coming and going from Italy. And they're bragging about what a wonderful time they had there. For the rest of your life you will say "Yes, that's where I was supposed to go. That's what I had planned".

And the pain of that will never, ever go away because the loss of a dream is a very significant loss.

But if you spend your life mourning the fact that you didn't get to Italy, you may never be free to enjoy the very special, very lovely things about Holland.

Emily Pearl Kingsley

The Genetic Support Networks

AGSA, The Association of Genetic Support of Australasia was established in 1988 and funding was received from the NSW Health Department in 1994 resulting in the employment of the present coordinator.

The Genetic Support Network of Victoria was established in 1997 through the Victorian Clinical Genetics Services as a result of recommendations from "Genetic Services in Victoria - A Discussion Paper" produced by the Department of Human Services.

The Genetic Support Coordinators, Dianne Petrie in NSW and Caroline Bowditch in Victoria, act as a convenient reference point for first contact between either a doctor or a family, and a specific national or state support group. Many people in other states have registered with AGSA and the Victorian families welcomed the establishment of the network in Melbourne. The Networks not only ensure that those with a genetic disorder have appropriate and accurate information and support to enable them to manage the challenges to their health and well-being, but also act as an advocacy group with local government. Both organisations believe that collaborations between individual, families, general practitioners and other health care professionals, and the genetic services is the essential basis for achieving this goal.

Genetic Support Network of Victoria:

Caroline Bowditch, Coordinator, Victoria is based at The Murdoch Institute, Phone (03) 8341 6315, Fax (03) 83341 6398, Email bowditcc@cryptic.rch.unimelb.edu.au.

Something to Remember....

Although there are many benefits in support groups, they are not for everyone! It is important that contact information for the relevant support groups be passed on to patients but they are allowed to contact them when the time is right for them - it will be different for each family. Participation in a support group can be very significant, and most people welcome it at some point, but not always at the earliest possible moment. We want to ensure that people are not

harmed from the experience, and that each is given a choice whether to get involved or not.

We recognise that support groups have a very different purpose to that of professional carers in a patient's life. They could be viewed as a part of the 'treating' team but they will never replace it!

Authors: *Caroline Bowditch, Co-ordinator,
Genetic Support Network of Victoria
Murdoch Institute
and
Margaret Sahhar
Senior Social Worker
Victorian Clinical Genetics Service*

CONFERENCES

Autism Association of NSW

Thursday Evening Programme

21st October 1999

Time: 6.30 pm - 8.30 pm

Guest Speaker: Temple Grandin

Cost: Family Member \$40

Professional \$60

Friday Day Programme

22nd October 1999

Time: 9.00 am - 4.00 pm

Guest Speaker: Temple Grandin

Venue for both Events: Australia Technology Park Conference Centre, Garden Street, Redfern,

Please send Registration to PO Box 361 Forestville 2087

For Enquiries contact Chris O'Connell-Gray

Phone (02) 9452 5088 or Fax (02) 9451 3447

Loretta Giorcelli, Anthony Warren and Parent Panel present "Children with Autism Spectrum Disorder in the Mainstream Classroom"

Cost: Family Member \$60, Professional \$100

PAM OWBIDGE
21 ELLERDALE STREET
STRATHPINE QLD 4500
PHONE 07 3205 3201
FAX 07 3205 3303

10th AUGUST 1999

TO WHOM IT MAY CONCERN,

LAST YEAR I PUBLISHED A PREMATURE BABY DIARY FOR PARENTS WHOSE BABYS' NEED TO STAY IN HOSPITAL AND THEY CAN RECORD ON A DAY TO DAY BASIS THEIR BABYS PROGRESS WHILST IN HOSPITAL. THE DIARY IS THE FIRST OF ITS KIND IN THE WORLD. IT HAS MEDICAL TERMS USED IN THE INTENSIVE CARE NURSERIES PUT INTO LAYMANS TERMS AS WELL AS KEEPSAKE PAGES.

THE DIARY IS SELLING IN ALL THE MAJOR HOSPITALS THROUGHOUT AUSTRALIA AND WE ARE ON THE INTERNET. THE HOSPITALS IN AUSTRALIA SELL THE BOOKS FOR \$20.00 AND \$5.00 OF THAT SALE GOES TOWARDS RESEARCH FOR PREMATURE BABIES. I SELL THEM FROM HOME MYSELF BY WORD OF MOUTH OR ADVERTISING IN VARIOUS MAGAZINES.

I AM JUST ABOUT TO GO INTO REPRINTS AND WOULD LIKE SPONSORSHIP OR SUPPORT FROM YOUR COMPANY/ORGANIZATION TO HELP ME OUT.

- 1) I CAN OFFER SPACE FOR ADVERTISING OF YOUR COMPANY AND OR PRODUCTS
- 2) I CAN OFFER SPACE LISTING DETAILS OF YOUR ORGANIZATION AND WHAT YOU DO TO OFFER PARENTS FOR SUPPORT
- 3)OR ANY IDEAS WHICH YOU MAY HAVE FOR YOUR OWN COMPANY/ORGANIZATION I WOULD GLADLY TALK TO YOU ABOUT.

IF YOU ARE INTERESTED IN ADVERTISING OR SPONSORING IN A ONE OF A KIND UNIQUE KEEPSAKE PREMATURE BABY DIARY IN THE WORLD PLEASE FILL OUT BOTTOM SECTION AND FAX BACK TO ME. THE REPRINTS WILL HAVE TO BE DONE FAIRLY QUICKLY AS HOSPITALS ARE ORDERING ALL THE TIME.

.....
NAME OF COMPANY/ORGANIZATION.....
ADDRESS.....
STATE..... POSTCODE..... PHONE..... FAX.....
CONTACT NAME..... POSITION HELD.....
WE ARE INTERESTED IN ADVERTISING IN DIARY.....
WE ARE INTERESTED IN SPONSORING THE DIARY.....
WE WOULD LIKE MORE INFORMATION PLEASE PHONE US.....

SIGNATURE.....

PRINT NAME.....

PREMATURE BABY DIARY - ONE DAY AT A TIME

PAM OWBRIDGE

NOTICE OF ANNUAL GENERAL MEETING

All members and friends of

AGSA

The Association of Genetic Support of Australasia Inc.

are cordially invited to attend.

The Annual General Meeting will be held at

66 Albion Street, Surry Hills NSW 2010

on

Sunday 17th October 1999

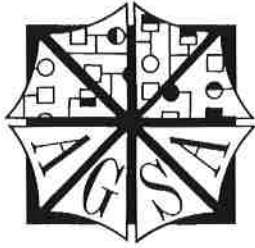
commencing at 10 am

The following items are attached for your information:

- * Agenda
- * Nomination Form
- * Proxy Form

Members are reminded that only current financial members are eligible to vote at the Annual General Meeting but this does not preclude attendance.

Ros Smith
President
29th July 1998



AGSA

THE ASSOCIATION OF GENETIC SUPPORT OF AUSTRALASIA, Inc

Funded by the NSW Health Department | Reg Charity No. CFN 15481

Tax ref: AF1595C/SF8566

66 Albion St, Surry Hills NSW 2010

Ph: (02) 9211 1462

Fax: (02) 9211 8077

Annual General Meeting

Sunday, 17th October 1999

at 66 Albion St, Surry Hills (Old Children's Court Building)

Time: 10 am for 10.15 am

AGENDA

1. Welcome and Introduction: Marlene Brightwell
2. Apologies
3. Guest Speaker: *to be advised*
4. President's Report: Marlene Brightwell
5. Information Officer's Report: Dianne Petrie
6. Treasurer's Report and audited accounts: Pam Bingham
7. Election of Office Bearers for 1999/00
 - a) All positions declared vacant and appointment of returning officer
 - b) Election.
The committee, including executive, comprises a maximum of 12 members.
8. Appointment of Public Officer
9. Appointment of Auditor
10. General business
11. Close of meeting

A light meal will be provided following the conclusion of the meeting.

Form of Appointment of Proxy
Rule 33(2)

I,.....
(full name)

of
(address)

being a member of the Association of Genetic Support of Australasia,

Inc., hereby appoint
(full name of proxy)

of.....
(address)

being a member of that incorporated association, as my proxy to vote for me on my behalf at the Annual General Meeting of the Association to be held on Sunday the seventeen day of October 1999.

.....
Signature of member appointing proxy

.....
Date

ELECTION OF OFFICE BEARERS FOR 1998/9

NOMINATION FORM

NB Only financial members at the time of the AGM are eligible to vote and/or hold office

Iam willing to accept
nomination for the position ofwithin the
Association of Genetic Support of Australasia, Inc.

Signed:

Date:

Proposed by:

Signed:

Date:

Seconded by:

Signed:

Date:

AGSA SUPPORT GROUP MEMBERS as at February 1999

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.
Coorinda Family Support Group
Cleft Pals, The Cleft Palate & Lip Society
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 Assoc of ACT
Cystic Fibrosis Foundation, North Ryde.
Early Education Clinic, North Sydney
DIAL (Qld)
Donor Conception Support Group
D.E.B.R.A.
Depressive & Manic Depressive Assoc.
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)
I.D.E.A.S. Inc
Klinefelter Syndrome Support Group
Kurrajong Early Intervention
Haemochromatosis Information Service & Support Group NSW
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)
Lysosomal Storage Disorders
M.P.S. Society
Marfan Syndrome Support Assoc. NSW
Meniere's (NSW) Support Group
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
Retinitis Pigmentosa Society of NSW Inc.
Rett Syndrome Assoc. of Aust.
Royal Blind Society of NSW
R.T.M.D.C. (U.K.)
SAFDA
SANDS
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 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 450 genetic conditions representing families and individuals seeking contact.)

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

**66 Albion Street
SURRY HILLS
New South Wales 2010
AUSTRALIA**

**Tel: + 61 2 9211 1462
Fax: + 61 2 9211 8077**

**Peer Support/Information
Officer:**

Dianne Petrie

Office Hours: 9.00 am - 3.00 pm
Monday - Friday

**Medical and Professional Advisory
Board**

Dr K. Barlow-Stewart
PhD; BSc
Prof. D. Sillence
MB BS; MD (Melb; FRACP; FRCPA, FAFPHM
Prof. G Morgan
MB BS; FRACP
Dr B Wilcken
MB; ChB; FRACP
Prof. R. Trent
PhD; BSc (Med); MB BS (Syd; BPhil (Oxon),
FRACP; FRCPA.

ANNUAL SUBSCRIPTION

Individual	\$20.00
Group/Organisation	\$40.00

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 **