

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

66 Albion Street, Surry Hills,
NSW 2010 Australia
Tel: +61 2 9211 1462

NEWSLETTER

APRIL 2000

ISSUE 47

MISSION STATEMENT

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

Reg Charity CFN154891 Tax Ref AF1595C/SF8566

The Registered Domain Name is now:

website: www.agsa-geneticsupport.org.au

Email: agsa@ozemail.com.au

Contents:

Editorial

Contact Corner:

**48XXYY, Familial Articulate Hypomobility
Type A, Chromosome 1p36del,
Glycogen Storage Disease Type 5 or
McArdle's Disease.
Genetics Disorders Awareness Week**

NOTICE

*This newsletter is brief because of financial
restrictions. This will not affect future
newsletters.*



POWERHOUSE MUSEUM,

***GENETIC DISORDERS AWARENESS
WEEK LAUNCH***

14TH JUNE 2000 AT 6.00 PM

***WE LOOK FORWARD TO SEEING
YOU THERE.***

ALL ARE WELCOME

EDITORIAL

Once again, join AGSA in launching **Genetic Disorders Awareness Week for Australia at The Powerhouse Museum, Ultimo, Sydney on 14th June 2000**. Awareness Week is a time to educate and to inform the public about genetic disorders. It is a time to recognise the work individuals and support groups do to help those affected. It is a time to create awareness, a time to further educate ourselves and others about genetic disorders. How many more rare genetic disorders will technology diagnose this year? For these people, AGSA will be there to offer information, support and contact. Where would the families be without a contact point and the many Support Groups under the AGSA umbrella? This year, please join us for an evening where both key speakers, Associate Professor Kathy North and Stuart Purvis-Smith, leaders in their own respective fields of genetic research and cytogenetics, will open your mind to a maze of new questions and answers. Also, be there for the announcement and to congratulate the NSW High School winners of the NSW Genetics Education Program Essay Writing and Poster Competition. Support AGSA and be a winner in their raffle. It will be a great night!

Raffle prizes include Westfield Shopping Plaza Gift Vouchers, Videos from Fox Home Entertainment, Golf Sports Shirts, Sparkling wine and more. Tickets are \$1.00 each or 3 for \$2.00 and are for sale on the night or prior to, by sending a cheque to the office.

The program, invitation and raffle information are enclosed in this newsletter. Our members, their guests and the general public are welcome to attend. Drinks and finger food are included in the price. Need more information? Please contact the office.

Marlene Brightwell
AGSA Committee

FUNDING CRISIS!!!!

We need your help! We need your help to insure AGSA can provide the level of service our clients need. AGSA has not received any increase in funding (other than the CPI index) from the Government for **6 years**. Our call rate and client base increases every year. We need to employ two full-time assistants in the office to carry this work load. Letters to Government Departments for enhancement funding have been unsuccessful. Again, we will be writing to the Minister of Health, Craig Knowles. AGSA is a peak organisation for Genetic Disorders providing support and contact and is a member of the NSW Genetics Advisory Board.

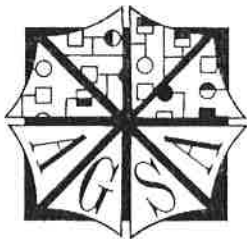
We need to see an increase in funding **immediately** so we can continue to provide the contact and information requested from our many clients and genetic support groups.

Ways you can help AGSA:

- **Write** in support of AGSA. to:
The Hon. Craig Knowles, Minister for Health,
Level 33, Governor Macquarie Tower,
1 Farrer Place, Sydney,
- **Lobby** your Local Government Representative. If you need AGSA pamphlets, please ring the office and they will be forwarded to you.
- **Pay** membership dues promptly for the 2000/2001 financial year (see enclosed).
- **Find** sponsors for us
- **Give** a Donation (\$2.00 and over are tax-deductable)
- **Hold** a fundraising event for us
- **Attend** Genetic Disorders Awareness Launch
14th June 2000

Be proactive and help AGSA let the Government know we are a vital organisation for those affected directly and indirectly by genetic disorders, now and in the future.

AGSA Committee



AGSA

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MEMBERSHIP SUBSCRIPTION FORM

SUBSCRIPTION YEAR OPERATES 1ST JULY 2000 - 30TH JUNE 2001

SURNAME/ORGANISATION _____

GIVEN NAME/ORGANISATION _____

ADDRESS _____

_____ POSTCODE _____

PHONE NO. () _____

IF ORGANISATION, CONTACT PERSON _____

IF INDIVIDUAL, Please indicate your disorder _____

IF PROFESSIONAL MEMBERSHIP, Please indicate your profession _____

SIGNATURE _____

(It is understood this information will be kept on a mailing list)

PLEASE PRINT NAME OF SIGNATORY _____

FAMILY/INDIVIDUAL (including GST) \$22.00 ()

ORGANISATION/SUPPORT GROUP (including GST) \$44.00 ()

DONATION (Tax deductible over \$2) _____

RENEWAL () NEW SUBSCRIPTION ()

If experiencing financial hardship please contact Dianne Petrie (02) 9211 1462

Please make cheques payable to: AGSA

and mail to:

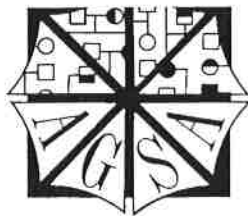
The Treasurer, AGSA

66 Albion Street

SURRY HILLS NSW 2010

AUSTRALIA

Date Received _____ Receipt No _____



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EHLERS-DANLOS WORKSHOP FOR PATIENTS AND AFFECTED FAMILIES

Sunday 21st May 2000, 10:00 – 3:00 p.m.

At AGSA, 66 Albion Street, Surry Hills NSW 2010

- 9.30 Registration – cost per person \$20.00
- 10:00 **The Ehlers-Danlos Syndrome – An Overview** David Sillence
- 10:30 **Repairing wounds with Tissue Glue** Kay Burns
(to be confirmed)
- 11:15 **Tests for people with Ehlers-Danlos Syndrome** David Sillence
- how they are performed and what they mean
- 12:00 **The Heart and blood vessels in Ehlers-Danlos Syndromes** _____
- 12:30 **Lunch**
- 1:30 **Management of Ehlers-Danlos Syndrome Brainstorming Session** Jenny Ault

Participants will be supplied with handouts which act as cues for discussion. We will use your participation to “brainstorm” solutions.

1. First Aid – bruises, cuts, sprains
2. School Issues – Preschool
3. School Issues – Primary
4. School Issues – High School
5. Workplace
6. Community Services
7. Specific Alerts / Emergencies

Ms. Kay Burns Clinical Educator – Accident & Emergency
New Children’s Hospital
Dr. Jenny Ault Dept of Rehabilitation, New Children’s Hospital
Prof. David Sillence Dept. of Clinical Genetics, New Children’s Hospital.

If you wish to attend please contact Dianne Petrie, AGSA, on 02 9211 1462, or send a cheque payable to AGSA and names of people wishing to attend. Cost includes registration, morning and afternoon tea plus lunch.



AGSA

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MAKING THE RIGHT CONNECTIONS SINCE 1988

Genetic Disorders Awareness Week

Wednesday 14th June 2000 at 6.00 pm

at

The Powerhouse Museum
Coles Theatre
Level 3, 500 Harris Street, Ultimo NSW 2007

Program

- 5.45 pm **Registration**
- 6.00 pm Cocktails and refreshments in foyer
- 6.55 pm **ENTER COLES THEATRE**
- 7.00 pm **Chairperson –**
Prof John Dwyer, Head of Medicine, Prince of Wales Hospital
- 7.10 pm Dianne Petrie - Peer Support/Information Officer, AGSA
- 7.20 pm Opened by – **Mr. Ian McManus MP**, Parliamentary Secretary for Health
- 7.30 pm **Mr Stuart Purvis-Smith – “From chromosome to Genes: Betwixt, Between & Beyond”**
- 8.00 pm **Living with Neurofibromatosis**
- 8.10 pm **Associate Professor Kathryn North – “What rare genetic disorders can teach us about common problems. The genetics of learning.”**
- 8.55 pm Discussion and question time
- 9.00 pm Prize Presentation to students by Dr Kris Barlow-Stewart, NSW Genetics Education Program
- Raffle Draw
- 9.05 pm Tea & Coffee in the foyer
- 9.15 pm Close

Kindly Sponsored by The Powerhouse Museum,

Transkaryotic Therapies Inc. USA and 20th Century Fox Home Entertainment

Parking is available at the Entertainment Centre, Darling Drive at a special rate (\$8.50). Bring the Parking Ticket with you to be stamped by the Powerhouse Museum. (Cheque payable to AGSA Post to AGSA, 66 Albion Street, Surry Hills NSW 2010)

RSVP and payment by 1st June 2000 Cost per person is \$20.00.

I will be attending Yes No (please circle) I enclose cheque for \$ _____
Name/Organisation _____
Address _____ Tel: _____
Condition (if applicable) _____
No. of people attending _____
Attached list of names please.



Funded by the NSW
Health Department

Creating awareness and providing contact, peer support and education to people affected by genetic disorders.

“MAKING THE RIGHT CONNECTIONS SINCE 1988”

Official Launch of

Genetic Disorders Awareness Week

Wednesday 14th June 2000 at 6.00 pm

at

The Powerhouse Museum

Coles Theatre

Level 3, 500 Harris Street, Ultimo NSW 2007

Speakers

Assoc. Prof. Kathryn North – *“What rare genetic disorders can teach us about? common problems. The genetics of learning.”*

Kathryn North is currently Associate Professor in Paediatrics at the University of Sydney, Head of the Neurogenetics Research Unit at New Children's Hospital, Westmead. Assoc. Prof. Kathryn North is the first recipient of the Career Research Development Award and winner of the 2000 Sunderland Award for outstanding Neurological Research.

Stuart Purvis-Smith – *“From Chromosomes to Genes: Betwixt, Between and Beyond.”*

Stuart Purvis-Smith is the Principle Scientist and Unit Manager of the newly created Molecular and Cytogenetics Unit in the South Eastern Area Laboratory Services (SEALS) at the Prince of Wales Hospital, Randwick.

Posters by students in Years 7-9 advertising Genetic Disorders Awareness Week and essays on ‘genetics in the future’ written by students in Years 10-12 will be on display. First Prizes will be presented on the night.

If you wish to attend please contact

Dianne Petrie:

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