



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

FUNDED BY THE NSW HEALTH DEPARTMENT

NEWSLETTER

FEBRUARY 2001 ISSUE 51

ISSN 1033 - 86524

MISSION STATEMENT

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

ABN 83 594 113 193

Contents

Contact Corner: Emery Driefus Muscular Dystrophy, Epstein's Anomaly of the Tricuspid valve, Jacobson syndrome, ATR-X Syndrome, Pompe's Disease Type 11, NF! And Noonan syndrome.

Profile: Canavan Disease

Media Releases – National Education

Evaluation, Genetic Privacy

Conferences

EDITORIAL

What happens to a child with a genetic condition or a disability for that matter, who is in a special education unit or a special school when they turn 18? I would like to hear from you about your experiences trying to find a suitable work place or education centre and to hear about your experiences with the Atlas Program or Post School Options as it used to be called. Why is it made so difficult? Does it all come down to lack of funding? I think the problem is far greater. Why isn't there a balance between a sheltered workshop and tertiary education? Suddenly after being in a protected environment they are meant to leave school, independently catch buses, find their way around education centres and if you are lucky, they may have an aid to assist them.

Available Programs are usually not nearby. I know of a young Down syndrome man who has to get up at 6.00am, catch two buses and a train to attend a cooking class at Petersham. The course costs \$3,000, which comes out of the \$13,000 allocated by the Atlas Program. He is on trial to see if he is suitable?? Shouldn't the courses be designed to be suitable to the person with an intellectual/physical disability?

What happens to the carers? Who helps them?

They have to once again, fill in detailed forms, attend expos to seek out disability centres, visit

the centres, make appointments, talk to teachers, fill in more forms. Meanwhile life is put on hold. Finally, when a suitable place is found it is often booked out and there is a waiting list.

Let me hear your experiences, what you have found good or bad and what advice you can give.

“The true measure of a civilized society is its willingness to assist its disadvantaged.”

Best wishes,



Dianne Petrie

If you think AGSA does a good job, please write a letter of support to:

The Hon. Craig Knowles,
Minister for Health,
Level 33, Governor Macquarie Tower,
1 Farrer Place, Sydney.
Many thanks to those who wrote previously.

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.

NF1 AND NOONAN SYNDROME

A mother of a 10 year old boy would like contact with others. Contact has been made with three children with both these conditions in Queensland. If you know of other families please contact AGSA for further details.

EMERY DRIEFUS MUSCULAR DYSTROPHY

A seventeen year old boy in Northern New South Wales would like contact with another family.

EPSTEIN'S ANOMALY OF THE TRICUSPID VALVE

Parents of a three-month-old baby boy would like contact with others. Please contact AGSA for details.

JACOBSON SYNDROME

A lady in Queensland with a six months old girl would like to set up a Jacobson syndrome support group. If you would like more details please contact AGSA

ATR-X SYNDROME

A mother of a 10 year old boy would like contact with another family with an affected individual. Reason for contact is

1. advice about testing location, cost
2. prognosis
3. availability of testing for carriers of ATR-X.

POMPE'S DISEASE TYPE 11

A 65-year-old lady would like contact and information. Pompe's Disease Support Group can be contacted on email: cookiezz@tpg.com.au



Support Groups News as at 16th February 2001

Parent to Parent (New Zealand) - 2000

SibKids is a place where you can meet other brothers and sisters of kids with special needs from across the world to share information and discuss issues of common interest. SibNet is the adult version of Sibkids for young adult brothers and sisters. The website was created by Don Meyer, Director of the Sibling Support Project at Children's Hospital and Medical Centre in Seattle, Washington.
<http://www.seattlechildrens.org/sibsupp/sibkids/htm>

Landau Kleffner Syndrome

Landau Kleffner Syndrome: A Parent's Guide -
This recently published book by Richard & Katherine Price is available for \$15.00 plus postage. The book is clearly written and has a wealth of information on this childhood neurological disorder to help parents. Also included is a section targeting available services and listing of professionals that may help families within Australia.
Phone: (02) 9482 8425

**South Eastern Sydney Area Health Service -
November 2000** has launched the 'Directory of Services for Women's Health 2000', which provides contact details for a wide range of women services in South Eastern Health area and to assist service providers to make appropriate referrals. To order a copy of the directory, contact Kathy Petraccato on 9382 8158 / 9382 8280

Australian Sturge-Weber Support Group - Reaching Out - Spring 2000

Researchers in Germany and the United States are studying brain tissue taken from people with Sturge-Weber Syndrome during surgery to learn more about SWS and how it causes neurological damage. They hope to be able to find some different gene expressions between normal brain tissue and that of SWS individuals. Preliminary

results of the studies show that there are some differences in some protein production and modification. Understandably the brain tissue is not easy to come by and the SW Foundation have put out a worldwide alert for possible donations.

For further information contact

(03) 9762 4630 or

website <http://www.sturge-weber.com>

Australasian Tuberous Sclerosis Society Inc - Reach Out - November 2000

Information fact sheets are available on TSC. To request fact sheets phone
(02) 9630 3147.

National Health and Medical Research Council (NHMRC)

A Consumer's Guide: Early Breast Cancer: A comprehensive guide to the diagnosis and treatment of early breast cancer. The detailed booklet is based on a review of all available Australian and international research. For copies of the booklet, contact your state Breast Cancer Council, Cancer Information Service, Breast Cancer Support Service or (02) 9334 1700.

National Breast Cancer Centre (NBCC)

All About Early Breast Cancer: This booklet provides information as well as practical information such as the range and availability of support services and how to access them. This is also available on audio cassette. For copies of the booklet or audio cassette, contact your state Breast Cancer Council, Cancer Information Service, Breast Cancer Support Service or (02) 9334 1700

Fabry's Support Group Inc - June 2000

Trials are now in progress at the Royal Melbourne Hospital for Enzyme Replacement Therapy.

Health Consumer's Council WA (Inc) - November/December 2000

The Health Consumer's Council
website - www.hcc-wa.asn.au

**Australian Thyroid Foundation Limited -
Thyroid News - December 2000**

Members of the Foundation have offered their time and support to speak to fellow thyroid patients. Telephone support contacts are available for: Hashimoto's Disease, Children & Hashimoto's, Thyroid Cancer, Thyroid Surgery, Male Thyroid Disease, Congenital Thyroidism, Hypothyroidism, Grave's Disease, Grave's & Pregnancy, Orbital Decompression & Ted, Radioactive Iodine. For further information contact (02) 9890 6962 (answering service) or fax (02) 9845 7287.

Glossary of Genetic Terms

<http://www.bis.med.jhmi.edu/Dan/DOE/prim6.html>

Advertisement

"Mothersafe"

Medications in Pregnancy and Lactation Service

A new service for the women of NSW based at the Royal Hospital for Women
Comprehensive counselling service for women and their healthcare providers concerned about exposures during pregnancy and lactation of:-
Prescription drugs, Over-the-counter medications, Street drugs, Infections, Radiation, Occupational exposures.

Telephone: 9382 6539 (Sydney Metropolitan)
1 800 647848 (Non-metropolitan area) Mon-Fri

**National Council on Intellectual Disability
and
Inclusion National**

Media Release

Wednesday, 7 February 2001

NATIONAL EDUCATION EVALUATION

We scored a D!

Australia scored 'D's on the major measures of inclusion of children with disabilities in mainstream classes. On international measures we scored slightly

better with an average 'C'. Any score below a 'C' was classified as a failure by the raters.

In the research, parents, teachers, principals and academics were asked to rate their State on 3 measures of inclusion based on Australian criteria and 7 ratings based on United Nations criteria. The results have been forwarded to the United Nations as part of their evaluation of the progress of educational inclusion of people with disabilities throughout the world.

These results of national research on inclusion are presented in a special issue of the Journal 'Interaction' which is published by the National Council on Intellectual Disability in association with Inclusion National. Research papers are included on the results for each State as well as interstate comparisons in a national report.

The research found that for physical inclusion state scores ranged from 'D-' (NSW and Queensland) to 'C+' (

(Tasmania). 'Physical inclusion' measured the support for the presence of children with disabilities in the regular classroom.

For Social Inclusion, scores ranged from an 'F' (Queensland) to 'C-' (Tasmania, Victoria and the ACT). Several respondents noted that the other children were very skilled at including children with disabilities, but schools and education systems were much less capable. 'Social Inclusion' measured the support for the social inclusion of children with disabilities by the schools and education system. All States had much less success with curricular inclusion, which is the adaptation of the curriculum to meet the needs of the children with disabilities. Here scores varied from 'D-' (ACT, NSW, SA and Qld) to 'C-' (Tasmania and Victoria).

In comparisons between the States, Tasmania and Victoria scored significantly better than most other states. Queensland and NSW were clearly experiencing the greatest difficulty in including children with disabilities in mainstream education.

Western Australia, while scoring poorly overall, was credited as having some examples of inclusion that were at the level of 'world's best practice'. The reason for their poor rating seemed to be that the inclusion was restricted to a very few individuals.

In addition to the research findings, the authors of the national report conducted a review of the literature on inclusion of children with disabilities in mainstream classes. They reported that the

comparative research of inclusive versus segregated education strongly supported full inclusion, even for children with significant intellectual and physical disabilities. The findings were also positive on the impact of inclusion on other children and on teacher time. However, the authors stressed that successful inclusion required support for teachers and schools.

The intention of the National Council on Intellectual Disability is to repeat the research in future years to measure progress of the States towards the inclusion of people with disabilities in mainstream education.

NCID is also involved in the preparation of a report to Australian Education Ministers on the extent of Disability Discrimination in Australian schools and how we need to address this significant abuse of human rights.

Contacts:

Editors : Darrell Wills, PLEDG (WA) Tel: (08) 97913455, 0419 955 012

Bob Jackson, Edith Cowan University (WA), Tel: (08) 94005659, 0417-992527

Contributors

Belinda Epstein-Frisch, Family Advocacy (NSW) Tel: (02) 9869 0866

Tim Loreman, and Dr Joanne Deppeler, Monash University (VIC), Tel: 03 9905 2884

Deborah Byrne, TASCID (TAS), Tel/Fax: 03 6228 1911

Advocacy Action (ACT), Jackie Ford, Tel: 02 6288 3847

Fiona Campbell & Sandi Seymour (QLD) (SA) Tel: (08) 8232 6200

NCID

Paul Cain, NCID Tel: (02) 6280 0875

Reference:

Interaction, Vol. 14, Issue 2 & 3, 2001. Report Card on Inclusive Education in Australia. National Council on Intellectual Disability (NCID).
Attorney-General

MEDIA RELEASE

The Hon. Daryl Williams AM QC MP

Minister for Health and Aged Care

The Hon. Dr Michael Woolridge MP

7 February 2001

GENETIC PRIVACY

Australians will be able to tell the Government how they want their genetic information protected at public hearings that will be undertaken under the terms of reference of a national inquiry.

The Australian Law Reform Commission and Australian Health Ethics Committee of the National Health and Medical Research

Council will start their inquiry into legal and ethical issues surrounding the protection of genetic information immediately, the Attorney-General, Daryl Williams, and the Minister for Health, Dr Michael Wooldridge, said today.

The broad-ranging inquiry will examine the complex and significant privacy, discrimination and ethical issues posed by advances in gene technology. The ALRC and AHEC will consider whether laws are needed to:

Protect the privacy of genetic samples and information;

Provide protection from inappropriate discriminatory use of genetic samples and information; and

Reflect the balance of ethical considerations relevant to the collection and use of such samples and information in Australia.

The terms of reference require the ALRC and AHEC to conduct extensive consultations before reporting to the Government on 30 June 2002. A discussion paper on the key issues will be released later this year.

Genetic information has many important scientific and medical applications which are of benefit to the community as a whole.

However, the Government shares the community's concerns that genetic information should be treated sensitively to prevent the potential for abuse or discrimination on the basis of actual or imputed genetic characteristics.

The terms of reference are attached.

Media contacts: Catherine Fitzpatrick (Mr. Williams) (02) 6277 7300 Serena Williams (Dr Wooldridge) (02) 6277 7220

COMMONWEALTH OF AUSTRALIA

Australian Law Reform Commission Act 1996

National Health and Medical Research Act 1992

PROTECTION OF HUMAN GENETIC INFORMATION

1. We, DARYL WILLIAMS, Attorney-General of Australia, and MICHAEL WOOLDRIDGE, Minister for Health and Aged

Care, having regard to—

the rapid advances in human genetic technology; and

the scientific and medical applications of human genetic information which are, or could be, of benefit to the Australian community; and

ethical concerns in relation to the collection, storage and use of human genetic samples and information; and

the potential for inappropriate use or application of human genetic samples and information; and

evidence of, and the potential for, use of human genetic information by a number of sectors including employment; health,

including medical research, pharmaceuticals and health administration; insurance and superannuation; and law

enforcement; and

emerging issues about the control of, ownership of, and intellectual property rights in relation to, human genetic samples

and information;

refer to the Australian Law Reform Commission and the Australian Health Ethics Committee of the National Health and Medical

Research Council for inquiry and report pursuant to subsection 20 (1) of the Australian Law Reform Commission Act 1996

and paragraph 35 (3) (c) of the National Health and Medical Research Act 1992 respectively, matters relating to —

(a) whether, and to what extent, a regulatory framework is required-

(i) to protect the privacy of human genetic samples and information; and

(ii) to provide protection from inappropriate discriminatory use of human genetic samples and information; and

(iii) to reflect the balance of ethical considerations relevant to the collection and uses of human genetic samples and information in Australia; and

(b) any related matter.

2. In performing their functions in relation to this reference, the Commission and the Australian Health Ethics Committee shall—

a. conduct this inquiry jointly; and

b. identify and consult with relevant stakeholders, including the Privacy Commissioner and the Human Rights and Equal Opportunity Commission, and ensure widespread public consultation; and

c. have regard to the following matters —

(i) the rapid advances in human genetic technology including progress of research towards the mapping of the

human genome; and

(ii) the scientific and medical applications of human genetic information which are, or could be, of benefit to the

Australian community; and

(iii) evidence of, and the potential for, the inappropriate use or application of human genetic information; and

(iv) the range of Australian ethical opinion as to which, if any, uses and applications of human genetic information

are ethically acceptable; and

(v) the global dimensions of issues relating to research, regulation and the protection of interests; and

(vi) any relevant existing or proposed international law and obligations; and

(vii) any relevant constitutional issues; and

(viii) any relevant existing or proposed Commonwealth legislation; and

(ix) the implications of the decision by Australian health ministers to develop a national health information network;

and

(x) developments in other jurisdictions, including legislative and other regulatory action; and

(xi) relevant research and discussion of human genetic information privacy and discrimination issues.

3. The Commission and the Australian Health Ethics Committee are to report to the Attorney-General and the Minister for

Health and Aged Care by 30 June 2002.

Dated 5 February 2001

Daryl Williams Michael Wooldridge
ATTORNEY-GENERAL MINISTER FOR
HEALTH AND AGED CARE

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.

The following excerpt is reprinted from the Canavan Foundation website.

CANAVAN DISEASE

This section highlight stories about Canavan disease that have appeared in the news media. A brief explanation follows regarding these articles dealing with the lawsuit against Miami Children's Hospital and Dr Rueben Matalon. The Canavan Foundation is not responsible for any errors that may appear in the news accounts

On October 30, 2000, the Canavan Foundation together with two other non-profit organizations -The National Tay-Sachs and Allied Diseases Association Inc. (NTSAD) and Dor Yeshorim --joined parents of children affected with Canavan disease in filing a lawsuit against Miami Children's Hospital (MCH) and Dr Reuben Matalon. The unprecedented lawsuit

alleges that Matalon and his employer MCH, secretly obtained a patent for the Canavan disease gene that was discovered using the genetic information and financial resources provided by Canavan families and organizations. In addition, MCH then began to charge royalties that limited the availability of testing for the disease.

The six-count lawsuit alleged breach of informed consent, breach of fiduciary duty, unjust enrichment, fraudulent concealment, conversion and misappropriation of trade secrets.

After more than 13 years collaborating with Dr Matalon, the Canavan families and organizations never thought their efforts would result in MCH patenting a gene that could prevent others from gaining access to the testing they had collectively work so hard to make possible

In the lawsuit, the Canavan families and organizations primarily seek injunctive relief to prevent Miami Children's Hospital from restricting access to prenatal and carrier testing for Canavan disease and from impeding research on finding a cure or therapies for Caravan disease through enforcement of its patent.

CONFERENCES

"Conflict Resolution", Understanding & Dealing with Conflict. Seminars are at various locations in Sydney from 12th - 16th March 2001 - Penrith, Chatswood, Ashfield, Liverpool, and Parramatta. Contact Educational & Behavioural Consultancy (08) 8288 7511

THE FIRST EVER NEUROMUSCULAR CONFERENCE

**to be held 7-9th November 2001
at Sky City in Auckland.**

The keynote speaker is Professor Alan E.H Emery from the United Kingdom.

For further information please contact:

Denise Ganley, Information Manager,
Muscular Dystrophy Association,
PO Box 16-238, Sandringham, Auckland,
Ph 09815-0247 ex 1, Fax 09 815-7260 or
email Denise@mda.org.nz

Alzheimer's Association National Conference
27-30th March 2001 at the National Convention
Centre, Canberra.
For details phone: (02) 6251 0675

Guardianship Tribunal Seminar

This free seminar is focused on the needs of family members, friends and carers of people with disabilities. The seminar is not for paid service providers and other professionals. Wednesday 14th March 2001, 10.30am -12.30pm at Parramatta Leagues Club, Parramatta. For further information regarding this and future seminars within NSW call the Tribunal on (02) 9555 9049, 9555 8500 or access the website: www.gt.nsw.gov.au

**ADHD in the third millennium
Perspectives for Australia
16,17 & 18th March 2001**

John Lowenthal Auditorium
Westmead Hospital, Darcy Road, Westmead
The Children's Hospital Education Research Institute (CHERI) in association with the University of Western Sydney (Nepean), University of Sydney and the NSW Department of Education and training is proud to present its sixth Hippocrates and Socrates Conference.
Ph 9845 3017 Fax: 9845 3082

**THE BATTEN DISEASE CONFERENCE
2001 IN AUSTRALIA**

8.30 am – 5pm Saturday 21st March and
Sunday 1st April 2001

“Dockside” Cockle Bay side of Darling Harbour, Sydney

Morning Tea, Lunch & Afternoon Tea – price to be advised.

Accommodation: The Parkroyal, Darling Harbour holds a group book – information to be supplied.

The Australian Chapter of Batten Disease Support & Research Association aims to provide families and friends affected by this disease with information, education and social interaction in various aspects of Batten Disease.

Guest speakers to include worldwide researchers and medical professionals.

Contacts: Vanessa Anderson Ph: (02) 4334 5785 Beth Dowry Ph: (02) 4261 1655

Batten Disease Support & Research Association
www.bdsrc.org.

5 Oakshurst Close, Dapto NSW 2530 Register Charity No CFN 14877 ABN 56056118994 Email: gvicando@ozemail.com.au

We are happy to announce the 2001 Battens Conference will be taking place end of March at Darling Harbour, Sydney.

The mission of this Conference is to provide families and friends affected by Battens Disease or alike with the opportunity together at a central location in a friendly environment. To provide time for social interaction and information, training, and education in various aspects of this disease.

Our BDSRA USA groups have organized for worldwide researchers to share their knowledge updating us on research over the two day Conference duration.

**We would like to hear from any persons (family, friends, medical professionals) interested in attending, to assist us with booking numbers at the venue.

** Sponsors and/or donators are sought to assist families to travel from distances around Australia and New Zealand. Financial assistance will aid nursing and carer requirements.

**Medical and field specialist guests will be approached to speak on issues that are part and parcel of the disorders.

**If you assist caring for the affected kiddies and their siblings please phone Vanessa on (02) 4333 5785.

THE CHROMOSOME 18 REGISTRY & RESEARCH SOCIETY (AUSTRALIA) is currently organizing a conference 22nd – 23rd September 2001 in Sydney. For further enquiries please contact (02) 9580-5707

What's on?

Monday June 18th 2001 9.30 am – 3.00pm

Genetics in the Classroom: A collaborative seminar for teachers and parents

NSW Genetic Education Program and AGSA

Cost \$35 for parents and support group members \$75 for teachers. More details to following the April Newsletter
Enquiries

NSW Genetic Education Program (020 9926 7324

Correction to telephone numbers (02) should read (03)

December Newsletter Issue 50

Metabolic Dietary Disorders Association (MDDA)

Kerri Carboon (03) 9728 5510

Natalie Wiggins (03) 9850 7484

AGSA would like to apologies for any inconvenience.