



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

FUNDED BY THE NSW HEALTH DEPARTMENT

NEWSLETTER

**OCTOBER 1997 ISSUE 32**

**MISSION STATEMENT**

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

Reg Charity CFN15481 Tax Ref AF1595C/SF8566

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**PEER  
SUPPORT/INFORMATION  
OFFICER'S REPORT**

Thank you to the outgoing committee members for all your hard work and welcome to the new members: Ros Smith - President, Richard Petrie - Vice President, Dr Stephen Withers - Treasurer, Kerryn Allen, Jenny Blackwell, Sarah Bridge, Marlene Brightwell, Sharon Clark, Brenda Phillis, Debbie Redelman, Mandy O'Reilly, Louise Scott - Regional Representative and Lisa Smith. You will be able to read their profiles in the December issue.

On 19th October, AGSA held a Beckwith-Wiedemann Seminar with Guest Speaker, Dr Alison Colley, Clinical Geneticist from Liverpool Hospital. Twenty two people attended and all are eager to set up a support group. Caroline Hawes and Jo Zimmerman, the co-ordinators, are planning a picnic day for April/May 1998.

Due to the larger than anticipated response to the workshop on Fragile X on 8th November with Prof. R Hagerman and Prof Gillian Turner, the

venue was moved to Lecture Theatre 1, Sydney Children's Hospital beginning at 9.30 am.

The special XYY meeting with Dr John Graham Director, Clinical Genetics and Dysmorphology, Medical Genetic Birth Defects Centre, Cedars-Sinai Medical Centre, Los Angeles, will be held at AGSA on 21st November from 9.00 am till 12 noon.

Recently, I attended a one day workshop on 'Helping People with Disabilities Deal with Grief and Loss' conducted by Vivienne Riches. The workshop highlighted the need for people with a developmental disability to be able to express their emotions of grief by using various strategies.

Keep in touch by sending in small articles on what your group is doing.

DIANNE PETRIE

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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 to 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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### CONE ROD DYSTROPHY

A mother with a seven year old girl would like contact with others. Phone AGSA for details.

\*\*\*\*\*

### 46XX del 11q 13q21

Contact has been requested for her four year old daughter. Contact AGSA for details.

\*\*\*\*\*

### 49XXXXY

A father would like contact for his two year old son with this condition. He knows of 38 in the world and has received a lot of information from a lady in America.

\*\*\*\*\*

### MITOCHONDRIAL MYOPATHY

A gentleman would like contact with somebody else with this condition plus any information that is available.

Please contact AGSA for details.

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### PROFILE:

**FAMILY FOCUS WHEN A CHILD HAS A DISABILITY** by Dr Enver Bajraszewski

*Reprinted with permission from Australian Doctor Issue 21 June 1996*

Many people, including doctors, have an initial feeling of dismay or even helplessness when they are faced with the fact a child has, or might have, a disability.

For doctors faced with any medical problem there is a strong focus at first on the possibility of

communication, education and independence, and the whole family will need support.

### **What is needed by families of children with disabilities?**

Families of children with disabilities need to:

- \* Understand the nature and effect of their child's disability and what can be done to help.
- \* Receive help which recognises and respects the individuality, value and complexity of each family member in achieving personal needs and goals. Help is needed in a wide range of areas - physical, intellectual emotional and social.
- \* Feel they have left no stone unturned trying to do the best for their child to be cured, helped or compensated. They may need to try a range of therapies including unconventional therapies.
- \* Feel in control of decision-making.
- \* Come to terms with the consequences of the disability.

Family adaptation results in positive effects including feelings of acceptance, achievement and satisfaction for all family members.

This adaptation is one of the major goals of early intervention.

### **What is early intervention?**

Essentially, early intervention is about getting the best out of the child with a disability by meeting the needs of the whole family in ways that are important and acceptable to the family.

In early intervention there is active collaboration with the family in problem identification, definition and solution, goal setting, advocacy for the child and helping families access the services which are available or can be organised.

All aspects of child development need to be addressed, especially:

- \* Physical and intellectual development. eg, treatment of hearing and vision disorders and management of epilepsy.

\* Emotional/social development by focus on the child's complete developmental needs eg, behaviour, self-esteem and maximal involvement in peer group activities.

\* Maximal independence from others for activities of daily living, eg, by decreasing the handicapping effects of disability, use of assistive devices to achieve goals such as mobility self care and learning.

\* Strengthening of family function in areas the family considers the most important to best meet its needs and aspirations. Eventual family adaptation is a major goal. The role of the family is crucial and what is good for the family as a whole is usually good for the child.

### **Who provides services?**

Early interventional services are provided by a wide range of organisations which have a varying degree of funding by and attachment to federal, state and local governments.

### **How are services provided?**

Early intervention practice varies from a focus on disabilities to a family centred approach where the family has control over which intervention services are used.

Family-centred early intervention is based on the belief that the family can function most effectively if it has control over all decision to do with early intervention. It has been a major recent advance in helping children with disabilities.

Parents should be given information about the most important aspects of child development and how these are relevant to their child.

Written family services plans and individual program plans summarise everything being done to help the family, and are offered by many services.

Copies are given to families to use as desired.

The Australian Early Intervention Association is working to enhance the quality of early intervention services and can provide information about local services.

a cure. Anything less is often felt to represent a failure or to be of little value.

For parents, when they are first told their child has a disability, feelings of shock and hopelessness/helplessness are common early or later reactions.

**Eventually these are replaced by strong feelings of wanting something active to be done as well as needing practical and emotional support.**

A wide range of early intervention services are available which are aimed at meeting much more than the need for cure and which can assist families considerably.

Early intervention programs are consistent with the principles of family medicine because they go beyond focusing on only the disability.

**They recognise the individuality of children with disabilities and their families and the need for specific help and information.**

Families usually actively seek help. The role of all early intervention services providers is to work with families to help them get the information and services they need.

Families need to make their own informed choices about which services they use.

It is important for families with a child with a disability to be given information about services available as early as possible, and be provided with appropriate support.

Early intervention should not be delayed until a medical diagnosis is certain and is appropriately begun when developmental delay is thought to be likely.

Doctors and others who see children should know of early intervention services and be prepared to discuss them with parents.

**What are consequences of developmental disabilities in young children?**

Disabilities in young children can slow or prevent development of some or all of the following:

- \* Sitting, standing, working and running.
- \* Hand function and activities of daily living.
- \* Thinking, problem solving and learning survival skills.
- \* Understanding language, speaking and communicating.
- \* Vision and hearing
- \* Emotional growth and maturity: fulfilment, self-esteem.
- \* Social maturity: behaviour, independence and relationship.

In addition, there may be effects on the family which involve almost all aspects of family life, such as problems with child rearing, expenses and time commitments.

The family's reaction to hearing bad news (eg. denial and anger) and later problems with adaptation can produce enormous stress.

Burdens on the family can impact strongly on a child's development and must be addressed if the child is to make maximal gains.

The consequences of developmental delays for the child can range in severity from minor to devastating.

For example:

- \* Poor vision may be corrected with glasses: deafness caused by glue ear may resolve completely.
- \* Mild developmental delay may be normal variation.

But it may be due to intellectual disability and have a significant long-term impact on the child and family.

- \* Severe intellectual disability will cause problems with all areas of life, especially

For further information, telephone:

\* Hospital departments of child development/rehabilitation/general paediatrics.

\* State/local government departments of health/community services

\* Australian Early Intervention Association state chapters:

ACT (026) 205 6498

NSW (02) 9809 7055

NT (08) 89271 166

SA (08) 8243 8280

Tasmania (0362) 348 238

Victoria (039) 854 3353

WA (089) 443 0211

Queensland (073) 286 4262

\* *Dr Bajraszewski is a Paediatrician at the Department of Child Development and Rehabilitation, Royal Children's Hospital, Melbourne.*

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#### Did you Know...?

**People with disabilities and older people form a significant proportion of the NSW population. Approximately 25% of all people in the State have either a disability, are aged 60 or over, or are older with a disability. In NSW, 17% of the population has a disability, 13% are affected by a handicap, while 16% are aged 60 or over. (From *ADDING Connections, Aging & Disability Department*)**

Excerpt from

Charities Bulletin September 1997

#### Banking of funds

In the course of a fundraising appeal funds must be banked immediately. The word "immediately" means "without delay" or "without lapse of time". It is prudent for authority holders to employ a practice that ensure the immediate banking any proceeds. The practice must be practicable and appropriate, but provide appropriate standards of accountability. For example, the formulation of the practice should consider the amount of money involved and the location of banking facilities.

## CONFERENCES

### International Day of People with a Disability

3 December 1997

**It is a day to raise awareness of disability in our community, and to celebrate the achievements of people with disabilities and their contributions to local communities worldwide.**

**For further information:-  
<http://www.health.gov.au/ood>**

**Adelaide to host**

### DISABILITY BIENNIAL

1 -10 MAY 1998

**A national performing arts festival, an access conference and a national conference on spirituality are just some of the events that will make up the inaugural Disability Biennial.**

The SPARC Disability Foundation is facilitating an inaugural national arts festival to celebrate the creative talents and abilities of people with a disability.

### Media Release

#### May 1998 - Focus on Disability

Individuals and organisations are invited to be a part of the Adelaide Disability Biennial - an event that will include a performing arts festival, three national conferences, an exhibition and a number of seminars. All events will be held 1-10 May 1998.

**Arts in Action** will launch the Biennial with High Beam, Australia's first national performing Arts Festival for people with a disability. High Beam will be followed by three national conferences.

**Elegant Access** Solutions will look at access for people with a disability to housing, heritage buildings and streetscapes. The conference will include a combination of presentations, site visits and workshops. Guest speakers will include UK's Selwin Goldsmith, and Michael Fox the world president of the international access committee.

**Beyond the Ramp** will attract all those with an interest in spirituality. This event has already attracted key national and international presenters including US, university professor Dr Nancy Eiesland who has published a book on disability and the church. She will be joined by Fr Michael Lapsley who, as Chaplain to Africa's ANC during the apartheid period, became a target for a letter bomb attack which left him with severe physical disabilities.

**Work-Able** is a conference with a focus on practical outcomes. Each of the speakers and workshops will look at practical steps that organisation can take to assist jobseekers with a disability to find and retain open employment.

The Biennial will culminate in a three-day exhibition, ARLEX Adelaide that will appeal to people with an interest in equipment and services that assist people with a disability. Exhibitor enquiries are welcome.

Biennial Director, Jeff Heath, said that the advent will be the culmination of 18 months work by more than 20 organisations with an interest in disability issues. There is still time for other groups to register to be part of this inaugural event. Your group might hold an exhibit, a seminar, a statewide conference, or some other event.'

**Biennial information can be found on the web - [www.ability-network.com.au](http://www.ability-network.com.au).**

Contact Ability Network:

GPO Box 989

Adelaide SA 5001

Ph 08 8377 2295 Fax 08 8296 1688

Email: [linkmag@ozemail.com.au](mailto:linkmag@ozemail.com.au).

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## OCTOBER 1997      ISSUE 32 Central Coast Chapter of NALAG

**Invites you to hear**

***Dianne McKissock***

**Speak on 'Helping Children Grieve:  
Shared Family and Community  
Responses'**

**At the Central Coast Leagues Club, Gosford,**

**7.30 pm - 24th November 1997**

**For further information contact:**

**Liz Martin on 4369 1431**

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## **National Respite Conference**

**11 -13 November 1998**

**University of Melbourne**

**Call for Papers**

**Are you interested in presenting a paper or running a workshop at this important biennial conference. Your paper should focus on one or all of the conference themes and clearly address the interests of the people attending.**

***Conferences themes: Carers, Quality & Best Practice.***

**Who should attend or present a paper?**

Carers, services providers and policy makers with an interest in respite issues for:

- \* Families of children with a disability**
- \* Younger adults with a disability**
- \* The aged**

The conference will focus on innovative carer programs including the National Carer Action Plan, Futures for Young Adults and the Strategy for Carers.

**If you would like more information contact**

**Eileen Hayes**

**Association for Children with a Disability**

**590 Orrong Road**

**Armadale Victoria 3143**

**Ph: (003) 9500 1232 Freecall: 1800 654 013**

## **Changes to the NSW Public Health Act 1991 Concerning Reporting of Birth Defects to the NSW Health Department**

From 1 January 1998 in NSW, birth defects detected during pregnancy or in a child **under one year of age in NSW** will be notifiable to the NSW Health Department under the Public Health Act 1991.

Birth defects found when a baby is born have been notifiable since 1991 and are reported to the NSW Midwives Data Collection (MDC) in the Health Department. The NSW Birth Defects Register was established in 1990 and brings together information from the Midwives Collection and information provided voluntarily by families through doctors and hospitals.

Until now, the information provided voluntarily through doctors and hospitals has included the mother's and baby's initials and address. However, the NSW Birth Defects Register has had difficulty in matching up reports which come from different doctors and hospitals because sometimes the baby's name is not decided until some time after birth, and sometimes the baby's name changes. Also, pathology laboratories which do chromosome tests are not able to obtain consent from families and the reports currently sent to the Register do not have any information on name and address at all. It has not been possible to match these reports with other information on a mother or baby.

The changes to the law will allow information on full name and address to be collected so that the confidentiality of such information is explicitly protected by law. These details allow information for one person to be matched when information comes from more than one hospital or doctor. Paper forms are shredded and personal information, such as name and address, are removed from the computer database after five years.

The changes to the law will also mean that there will be more reliable information for planning of

health services, and for evaluating real and apparent clusters of birth defects.

Birth defects include structural malformations such as cleft lip and neural tube defects, and four medical conditions: cystic fibrosis, thalassaemia major, phenylketonuria and hypothyroidism. Congenital infections, cysts and tumours are not included.

When a report is made to the Birth Defects Register, a copy of the form will be given to the mother, or the parents in the case of a child. Information about the Register will also be provided. In the case of a child, it is recommended that the copy of the report form is kept in the Personal Health Record (Blue Book) as a permanent record for the family of what information has been sent to the Register.

Any enquiries about the Birth Defects Register may be made to Dr Lee Taylor in the Epidemiology and Surveillance Branch, NSW Health Department, telephone (02) 9391 9223 or facsimile (02) 9391 9232. The Manger of the Birth Defects Register is Ms Susan Travis who may be contacted on telephone (02) 9351 7747 or facsimile (02) 9351 7742.

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Q.4 Which of your medicines do you get as private prescriptions and which ones do you buy as over-the-counter products ? *(If you can't remember the name please describe what it is used for).*

*Private prescription*

*Over-the -counter*

Q.5 Are there any nutritional supplements and equipment you need to manage your genetic condition ? *(Please list them)*

Q.6 Do you get any of the medicines, nutritional supplements or equipment you need from the hospital ? If yes, please list them. *(If you can't remember the name please describe what it is used for).*

Q.7 Are there any medicines or nutritional supplements that you or your family would start to use, or would use differently, if they were available on the PBS ?  
(Circle your answer) **Yes** **No**

*If yes, please list what they are and how you would use them differently.*

Q.8 Have you ever reached the PBS Safety Net and been able to get your medicines free or at a reduced cost ?  
(Circle your answer) **Yes** **No**

If yes, how often has this happened and when did it last happen ? Which month and year ?

Q.9 There have been a lot of changes to the PBS in the last few years. How have these changes affected you and your household ?

Q.10 Is there any way the PBS could be changed to better meet your needs ? *(Circle your answer)*      **Yes**              **No**

If yes, please describe the changes you would like.

Q.11 When your doctor gives you a prescription, do you usually discuss the price of the medicine ? *(Circle your answer)*

**Yes**              **No**

How do you feel about this ?

Q.12 Some medicines are available under different brand names at different prices. Are the medicines you use the cheapest brand ? *(Please circle your answer)*

**Yes** .... all or most are the cheapest brand

**Yes**    some are the cheapest brand

**No**     none are the cheapest brand

I don't know or I'm not sure.

Can CHF contact you by phone to follow up on this information ?

If this is OK, please tell us your name and phone number (include the STD area code). This information will be kept **strictly confidential**.

Name ..... Phone (....).....

**(Thank you. Please removed the 3 pages, fold into 3 and post. Many thanks)**

**Association of Genetic  
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2 Norman Street,

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**ANNUAL SUBSCRIPTION**

Individual \$20.00

Group/Organisation \$40.00

*Subscription Year 1st July - 30th June*

*\* The views expressed in this Newsletter  
are not necessarily those of AGSA.*

**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.