THE ASSOCIATION OF GENETIC SUPPORT OF AUSTRALASIA INC.

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NEWSLETTER

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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

AGSA welcomes our committee members, Marlene Brightwell, Kim Palmer, Richard Petrie, Brenda Phillis and Mona Saleh. We look forward to working together and taking AGSA into the new Millennium.

Looking back over the years, AGSA on a very small budget, has achieved an enormous amount. We have run over thirty seminars on specific genetic conditions, the Contact Register has grown from 150 to over 500 conditions. We have continued production of the AGSA's bi-monthly newsletter and our membership has increased 150% in the last five years. Genetic Disorders Awareness Week is an Annual event and grows in importance and strength each year with more and more people participating. Many still do not understand the role of genetic clinics and genetic counselling and Genetic Disorders Awareness Week not only makes people aware of these services, it increases awareness of the many different genetic conditions.

A special thank you to all the professionals who continue to refer their patients to AGSA. In turn, AGSA is able to refer callers on to appropriate services, e.g. early education clinics, medical services, etc. and especially provide contact with another family. People who call AGSA are grateful for a number of reasons. Many callers ring because they are unsure of their diagnosis and of its implications and they need to talk it

over with someone outside the medical and family settings. AGSA can provide this anonymity. We urge you to hand our pamphlet to each new family.

AGSA breaks down the feeling of isolation after the diagnosis of a genetic condition and has a vital role in the grieving process.

"making the right connections since 1988"

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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ORAL FACIAL DIGITAL SYNDROME

A mother of a seven month old girl would like contact with another family.

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PSORIATIC ARTHRITIS

A lady with this condition would like to hear from another person.

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TAR SYNDROME

A family knows of five families with a child with this condition and would like to make contact with others.

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SPONDYLOCOSTAL DYSPLASIA plus AGENESIS of the CORPUS CALLOSUM

A mother with six year old girl would like to make contact with another family.

Chromosome 21 with a partial monosomy

The baby is 4/52 is not thriving and the parents wish to make contact with other parents who have experienced what they are going through.

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RESEARCH

'THE RETT GENE HAS BEEN FOUND'

The momentous words above were used by Kathy Hunter, President of the International Rett Syndrome Association (IRSA) to begin an email that she forwarded to RSAA on 1 October. She was referring to the discovery of what is believed to be the cause of Rett syndrome, namely, a defective gene call McCP2 (pronounced meek-pea-two) which is located on the X chromosome at Xq28.

On the same day that we received Kathy's correspondence, announcement of the discovery was made in joint medical releases issued by the Baylor College of Medicine, Houston and the Stanford University School of Medicine. It was the result of collaborative efforts of researchers at these two American institutions which led to the detection of mutations of the McCP2 gene. Publication of their findings appeared in the October 1999 edition of the scientific journal 'Nature Genetics".

"The Rett syndrome gene, McCP2, encodes a protein (McCP2) which is involved in one of the many biochemical switches that is needed to

control the complex expression patterns of other genes by telling them when to turn off. This 'housekeeping protein' is critical for brain development and essential for life itself. Scientists believe that lack of a properly functioning McCP2 protein could allow other genes to come on or stay on at inappropriate times in development, thus disturbing the precisely regulated pattern of development."

Whilst it is likely that the discovery will not have a practical impact on Rett syndrome individuals for some time, it does open significant opportunities for further research and experimentation. Hopefully, it will provide a biological marker for molecular diagnosis; be a means of pre-natal testing to detect the syndrome in families with an affected individual; be a test for sisters of persons with the syndrome; and prove to be a basis for developing strategies to prevent the disabling effects of the disorder.

A much more detailed report of the discovery will appear in the next edition of the RSAA newsletter which is expected to be released in mid November. However, in the mean time, you are welcome to contact me should you required any additional information

Yours sincerely,

Bill Callaghan
President
Rett Syndrome Association of Australia

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From the Alpha-1-Antitrypsin Research Project Newsletter Volume 1, Issue 7 Autumn 1999.

A1AT Research at St Vincent's

Current research in our Biochemistry laboratory is still focused on the levels of Secretory Leucocyte Proteinase Inhibitor (SLPI) in both blood and lavage fluid (taken from the lungs during a bronchoscopy).

A number of samples of lavage fluid (salty water squirted into the lungs which mixes with normal lung fluid and cells) have been taken from people undergoing routine bronchoscopy. The level of SLP1 has been measured to help provide a normal range of comparison with people who have an A1AT deficiency.

We have been very fortunate to have two kind volunteers who underwent a bronchoscopy so we could obtain a sample of their lavage fluid for comparison with a "Normal" population.

We are still awaiting the results as the tests are very specialised and need to be repeated a number of times.

We will need to obtain many more samples of lavage fluid in Alphas to provide a meaningful range of SLO1 levels for A1AT deficiency.

If you are interested in helping us with our research please call Sue Brenton on (03) 9288 3135.

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Online Genetics Glossary: The National Human Genome Research Institute (NHGRI) at the National Institutes of Health provides an online multimedia glossary appropriate for professionals and the general public. This glossary contains user-friendly, downloadable explanations of genetic terms. A limited number of CD versions are available to educators and libraries free of charge. **Visit - http://www.nhgri.hih.gov/DIR/VIP/Glossary**

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Report from Alliance of Genetic Support Groups.

The Alliance Travels Overseas (excerpt).....

The meeting of the European Alliance of Genetic Support Groups (EAGS) in Geneva, Switzerland, explored the possibility of organizing an international meeting of "Alliances" in the year 2001 at the International Society of Human Genetics in Vienna, Austria.

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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.

FAMILY STORY

The following article was published in The Sunday Times Magazine 11 July 1999 by Times Newspapers Ltd. 1999. I recommend this article to you for its simple statement of facts and their impact.

A LIFE IN THE DAY OF ELSIE CHRISTIAN

by Elsie Christian.

Most mornings are the same. Around 5.30 Keith will start humming. Well, it's more of a drone. The crashing of drawers and cupboards will follow. I slip into my dressing gown and head for his bedroom. He greets me with the same big smile, and for the millionth time he'll tell me he loves me - though the words come out more "ove'ou".

We moved here from an old terraced house four years ago. Mainly to give Keith more space because of his continual walking about the house. Here there's lots of room, though the carpets do get worn out and the door handles have been replaced twice because of his coming and going. In the summer he might venture into the garden and sit in the shade of the Crimson King maple, but he never stays more than 10 minutes before he's off walking around the house again.

I have the physique of Edith Piaf, so it not easy of a morning for me to bathe someone who is 6ft tall and weighs 14 stone. I tried him in the shower once, but he just stood there while the water rumbled over him and me, while I tried to help.

Sitting down to breakfast with Keith is like watching somebody who's been on a hunger strike for a month. He eats two slices of wholemeal toast with marmalade and a bowl of cereal, washed down with a mug of steaming hot tea, all before the caffeine from my coffee has kicked into my bloodstream. I try to look beyond Keith's disability and treat him as a normal person. But chastising him, as I have to do sometimes, hurts me more than it hurts him. I tell myself that he is lucky knowing nothing of wars or drugs. But then I think of how unlucky he is, because he'll never get married and have children.

His transport to his daycentre arrives at exactly 8am. I'll stand at the door to wave him off, then head back to the kitchen for another cup of caffeine. My other son, John, usually falls out of bed as Keith leaves, then hogs the bathroom until he's satisfied that he couldn't make himself any more handsome. He has girl trouble and works as a waiter in the local college restaurant.

Once I'm showered and changed, John will have left for work, and my husband is in the bathroom. He was made redundant when the local kitchen factory closed down. He's suffered with high blood pressure ever since. To keep myself sane I recently completed a course at the Liverpool University of Further Education, writing screenplays. I enjoyed it so much that I spent a few hours a week sitting at the local library, reading anything to do with writing for television. This was a perfect excuse to treat myself to a computer.

Have a daytime job is something I miss. Not for financial reasons, but just to be involved with other people. I dread to think that I could end up

walking the aisles in a supermarket just for the sake of conversation. I told all my friends that when Keith was born I would be back at my job as a dental receptionist. But it wasn't to be. He was born brain-damaged. We didn't know why then and we don't know why now. It's not something that Robert and I discuss any more, simply because Keith would still be brain-damaged even if we did know,

My lunch is a coffee and a sandwich with Robert. I talk frequently and with concern about what will happen to Keith when we're gone. I wouldn't expect John to take care of him, as it wouldn't be fair. Robert says I worry unnecessarily, but I know he worries too. I shouldn't really, because Keith goes to a wonderful respite centre called Holly Road. They have a dedicated staff who dish out lots of TLC. He stays there a few days in the year to give Robert and me a break. We usually take off; then for a weekend to our favourite place: Llandudno. The freedom is wonderful. Best of all, though, is being able to sleep in together - something we can't do at home. Then with fully recharged batteries, we return home to begin again.

Keith will become a resident of Holly Road when Robert and I are no longer here. I get upset thinking he would be waiting for me to come and pick him up. I suppose other parents in the same situation worry just as much. Someone told me once how brave I was. I couldn't understand what they meant. Were they saying it because I care for someone? Or because other people stare at him when we go out together? Bravery belongs to the persecuted. I take care of Keith because he's my son and I love him.

If I'm not struggling with the computer, Robert and I might take off for long walks. The local park has become our little piece of countryside, where we talk of retiring to a little cottage in the country, somewhere in Cornwall or Devon. But that's all it is: talk. We need to stay here because of the excellent services Keith gets. There's so much available here for people with learning difficulties.

At 4pm Keith returns from the centre. He'll run up the drive with outstretched arms and greet me with another "ove'ou", almost knocking me over. When tea's ready, we'll all sit round the dining-room table, eating something spicy. Usually it's a curry. We chatter to Keith, asking questions about his day, but we never get a response.

Bedtime is winding-down time. John will be out with his friends. Keith will be in bed asleep, clutching a Scooby Doo video, which will be retrieved from the floor at least four times through the night. Robert and I will sit together on the couch. We'll turn off the television and listen to the silence of the evening, which is broken occasionally with Keith's loud snoring.

On my way to bed I'll go quietly into his room to give him his goodnight kiss. I leave him to dream his dreams, whatever they are, then go to my room. I'll set the clock for 6am. My tired eyes will close away another day, and I fall asleep listening for him, should he need me. It's a talent I've mastered over the years.

CONFERENCES

2nd National Conference: *'Justice for everyone, Intellectual disability and the law'*

12 -13 November 1999

Wollongong University

More info: Jan May or Margaret Bowen Ph: (02) 4228 4500

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A VIDEO ON CHILDREN AND ADULTS WITH CRI DU CHAT

CRY OF THE CAT was produced, written and directed by Helen McGrath (a parent of a child with the syndrome). It addresses the lack of literature available to new parents to the syndrome, and guides them in the understanding of how their child may develop as they grow.

Using a positive approach, the video aims to show the range of the syndrome from mild through moderate to severe developmental delay.

Filmed over four years it follows the life of four Sydney Cri Du Chat children, with brief appearances of eleven other children from around Australia.

It looks at the prognosis, the impact of that prognosis, early intervention, schooling, therapy, speech, family and support networks and future. There are interviews with geneticists, educationalists and therapists.

The video is for parents, families and friends, doctors, teachers, therapists and anyone who deals with a Cri Du Chat child.

(A portion of funds received from the sale of this video goes to the Cri Du Chat Support Group Australia)

ORDER FORM

CRY OF THE CAT

Name _____

Address _____

_____

State _____ Post Code _____

Country _____

Phone _____

Fax _____

Email _____

Cost (AUS) $45 each

No of copies: _____

Total @ $45.00 each _____

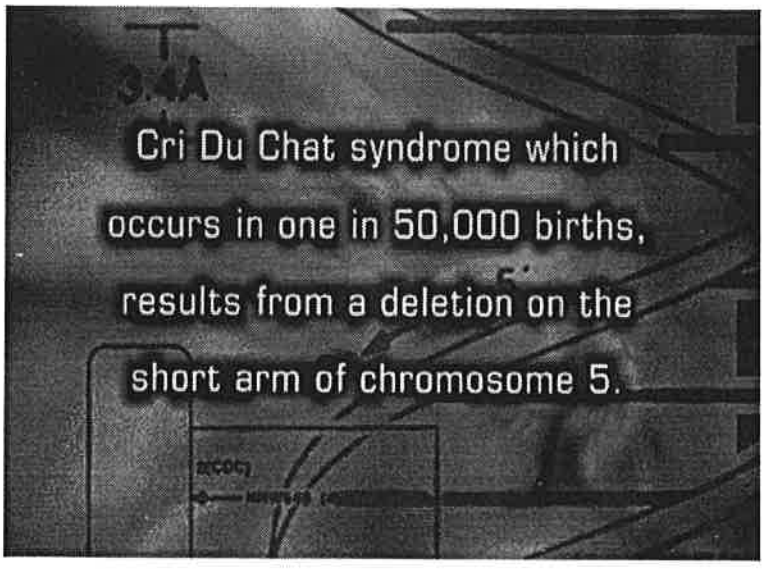
Plus postage $5.00 within Australia -
$13.00 outside Australia _____

plus $3.00 postage for each extra tape ordered

Total (AUS) $ _____

Please make bank cheque, in Australian dollars, payable to Helen McGrath.

Please post order form with cheque to:
Helen McGrath,
3 Browns Lane
North Sydney, 2060 Australia



MEDIA RELEASE
23rd August, 1999

$1.8 million for new Centre of Excellence in Adult Cystic Fibrosis Care

Cystic Fibrosis Victoria (CFV) has applauded the State Government's decision to build a $1.8 million specially-designed Centre of Excellence in Adult Cystic Fibrosis Care at The Alfred Hospital.

Construction of the purpose-built centre (to be located on the 5th Floor of the Alfred's main ward block) will begin early next year and take approximately eight months to complete.

CFV Chief Executive Officer Monica Walters said the Centre of Excellence will impact directly on the quality of life of the 240 adults in Victoria who live each day with Cystic Fibrosis, a life-threatening genetic disorder which severely attacks lung function and digestion.

The new centre will feature 33 beds in single and dual patient rooms, each with their own bathroom facilities. Eight isolation rooms will be specially-equipped with negative air pressure ventilation to reduce the chance of cross-infection which is especially important for those Cystic Fibrosis patients who have undergone a lung transplant.

"The new facility has been designed with patients' views in mind," Ms Walters said. "It will respect the need for patient privacy and exercise, it will help reduce the chances of cross-infection and those inappropriate experiences that can occur in larger shared ward arrangements."

The State Government's decision to fund the centre is a major success for the Victorian Cystic Fibrosis Care Alliance which was formed only 12 months ago.

The Alliance, of which Ms Walters is Convenor, comprises 10 leading health organisations which care for and treat the 600 Victorians who have Cystic Fibrosis. The organisations include Cystic Fibrosis Victoria, Royal Children's Hospital, Alfred Hospital, Monash Medical Centre, Victorian Clinical Genetic Service, Very Special Kids, Royal District Nursing Service and regional physicians.

"We couldn't be more delighted with the outcome of the Alliance's long and intensive lobbying to get this project off the ground," Ms Walters said. "The Minister for Health, Mr Robert Knowles, should be congratulated for approving the capital funding required for this much-needed facility."

Although there is no cure for Cystic Fibrosis at present, recent improvements in medical treatment and patient self-management have resulted in patients living longer and better quality, lives.

In the 1970s, many babies born with Cystic Fibrosis would only live until their teens; today, more than half will survive into their thirties and beyond.

For more information please contact: Monica Walters, CEO Cystic Fibrosis Victoria and Convenor Victorian Cystic Fibrosis Care Alliance. PH: 9686 1811 FX: 9686 3437 AH: 9826 2290.

DEPRESSION AND MOOD DISORDERS ASSOCIATION OF NSW

Subscriptions and postal enquiries: 41 Ilka St Lilyfield, NSW 2040
Information about current support groups: Mental Health Information Service 02 9816 5688
The DMDA is a Standing Committee of the NSW Association for Mental Health 62 Victoria Rd Gladesville, NSW 2111. Registered Charity CC15515 Donations over $2 are tax deductible

Dear member

Below is some information about a new treatment to reduce tobacco consumption. This is part of a research study at the University of Sydney and is part of some interesting new research into links between bipolar disorder and smoking. If you would like further information about this study please contact me on 02 9660 7413.

Kind regards

Meg Smith (President)

Treatment of Tobacco use in Bipolar Disorder

This is a study of a new cognitive behavioural treatment to help you reduce tobacco intake. This study will involve you and one other person. This could be a family member, a close friend, or even a health professional. You would choose who that person would be.

The treatment involved in this study will be in addition to other care you may be receiving from a health service or health care provider.

The sessions will be held within the University of Sydney or at other appropriate places.

The purpose of the study: The study is to assist you to reduce the amount of cigarettes you smoke by means of group treatment program.

The benefit to you: Do you think you are smoking too much? It is expected that this treatment program will benefit you by assisting you to reduce your smoking. The only stress/discomfort/inconvenience that could occur is in relation to reduction of the amount of nicotine you are taking in - and the treatment program has been specifically developed with this in mind. Any information you may give during the course of the program will be confidential. You have the right at any time to withdraw from the program.

Sessions: There will be one hourly group sessions over a period of about three months looking at simple techniques for monitoring tobacco intake. We will work together to develop ways to help you to reduce your tobacco intake and develop additional skills and techniques (for example alternative ways of problem solving and relaxation training)

We look forward to having you as a participant in our project.

Project team: M/s Dianne Clark, Assoc. Prof. David Kavanagh, Prof. Stephen Touyz

To take part in the study please contact: **Dianne Clark Dept of Psychology University of Sydney 9351 7523**

Dianne Clark ph.: 02 9971 2355 or write to 69 Rose Ave Wheeler Heights 2097.

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Monday - Thursday
Closed Friday

ANNUAL SUBSCRIPTION

Individual	$20.00
Group/Organisation	$40.00

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.

* 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****