THE · ASSOCIATION
OF · GENETIC · SUPPORT
OF · AUSTRALASIA

FUNDED BY THE NSW HEALTH DEPARTMENT

JUNE 1995 ISSUE 18

MISSION STATEMENT

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

Registered Charity No.C.C.27702

EDITORIAL

For most of us, Australia-wide, June conjures up images of tax returns and their associated problems; finding that missing receipt or the need to recap on the year-to-date and forward planning are essential pre-requisites to running a household, business or support group. Although some months short of our AGM (November) I would encourage all members to "forward plan" and consider nominating for a position on the AGSA Management Committee for 95/96. A number of executive and committee positions will become vacant and it is essential that we receive nominations from support group personnel, individuals, families and medical and allied health professionals so that representation is fair and just.

A decision is not required at this stage but some thought about how you might contribute or be involved could prove invaluable when AGM nominations are forwarded. Should you wish to know more about the role and responsibility of committee positions, please do not hesitate to contact me on (047) 515 872.

Trivia Night - 6th May 1995 (Charity Awareness Week)

To those who attended the Trivia Night I would like to express my very sincere thanks. It is often difficult juggling the fundraising needs of an individual organisation such as AGSA against the very real need for funds within your own groups.

ISSN 1033-8624

The evening was a great success, due largely, to the efforts of Margot Latham (Treasurer) and her friends, who provided the expertise. Many great prizes were donated, including an accommodation/ meal package from the Waratah Central Hotel, won, I might add by an AGSA Committee Member, a dinner set from Epping Day and Night Pharmacy, sporting goods from Sports Leisure and others too numerous to mention.

NSW Genetic Education Program-
No doubt many of you will be disappointed to learn that Amanda O'Reilly has resigned her position with the NSW Genetic Education Program. Mandy acted as the liaison person between the Education Program and AGSA over many years. Her own personal experience of having a child with a degenerative neurological disorder, and her involvement with support groups, has brought about an awareness of the issues surrounding families in crisis and an ability to deal sensitively with those concerned. I am sure you will all join with me in wishing Mandy well in her new position.

ROS SMITH
PRESIDENT

****

SUPPORT/ INFORMATION OFFICER REPORT

The following are some comments I feel are important and they will serve as a reminder for our Living Grief Seminar.

"I denied my own grief
Trying to be strong
It drove me crazy"

"Grief is your friend
It lets you remember
It lets you mourn".

Often in looking after everyone else we forget ourselves - this seminar is a time for you to stop and remember.

The profile this month is on mental illness research and depression. Having a disability or being the parent of a child with a disability is an on-going struggle to accept and live with the condition and lead a normal life. The extra burden of medication, teaching, dressing, feeding, worrying - there is always something to watch out for or something to fight for and sometimes the responsibility is overwhelming. This newsletter is a recognition of the struggle and acknowledges the fact that no matter how special a child with a rare genetic disorder may be, or no matter how strong and capable a person with a genetic condition may be, universally we would wish it otherwise.

Just a reminder, there are supports groups in NSW for the following:-

Klinefelter Syndrome
Leigh Disease
Rare Chromosome Disorders
Sotos Syndrome - addresses can be obtained by ringing me.

Margaret Rankin and Lance McMillan are holding a Haemochromatosis meeting at AGSA on 20th August 1-5 p.m. The guest speaker will be Dr. Mark Bassett. (see insert for details).Hope to see you there.

On 16th September 1995 AGSA goes to the regional area of WAGGA for our first rural genetic support seminar. Please notify members in that area to mark it in their diaries.

Thank you to all those people who ventured out on such a wet and miserable night to attend our second Trivia Night. $580.00 was raised which is fantastic. Thank you Margot Latham, the committee and friends.

I am having the second week off in the July school holidays and calls to the office will be answered by the committee.

Best wishes

DIANNE PETRIE

PROFILE A - Z GENETIC CONDITIONS

It is the intention of AGSA to profile, in each issue, a particular Support Group/Disorder, through 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.

For your information we profile.....

PSYCHIATRY AND GENETICS

THE SEARCH FOR THE MENTAL ILLNESS GENE(S)

Dr.John Collins

Over the last decade, articles describing the identification of linkage markers, and indeed the genes responsible for diseases have been very common in journals. Fields as diverse as haematology and cancer research have been transformed.

However, the mode of transmission of the common major mental illnesses such as schizophrenia and manic-depressive disorder is obviously quite different from that of say cystic fibrosis and other single gene disorders. Nevertheless, the situation is not so different from that in other common disorders with a strong genetic component such as coronary heart disease or diabetes which do not share single Mendelian patterns of inheritance. Unfortunately, to conceptualise schizophrenia as a multifactorial polygenic disorder does not provide any biological insight into the nature and number of genes involved or their mechanism of action.

Coronary heart disease, the multifactorial disease in which most progress has been made, provides a model of how genetic variance in such a disorder can be broken into its constituent parts. One of the first components to be characterised was familial hypercholesterolaemia --

individuals heterozygous for the causal gene have very high levels of circulating low-density lipoprotein and in consequence are prone to heart attacks early in adult life. Although this gene devastates the affected families, it still only accounts for a small proportion of heart attacks in the population. However, the other of the "polygenes" of this multifactorial disorder are gradually being identified.

Alzheimer disease was the first major psychiatric disease to attract attention. The interest was aroused by the observation that individuals with Down syndrome who survive into early middle age develop a dementia whose neuropathology is identical to that of Alzheimer's disease. The fact that chromosome 21 is triplicated in Down syndrome provided a strong clue that a mutual gene for Alzheimer's disease might be sited there. Further excitement followed the demonstration that the gene for beta amyloid found in the plaques of Alzheimer disease patients was in this region and this was proposed as a "candidate" (or causal gene). However it was soon shown that Alzheimer disease did not segregate exclusively with one allele for the amyloid gene.

In respect of manic depressive illness a few families have apparently yielded linkage with X - chromosome markers. Linkage has been claimed with colour blindness, G6PD deficiency and the factor IX gene. Totally different evidence of linkage of this disorder and two markers on the tip of short arm of chromosome ll came from a study of "Old" Order Amish, a religious community in Pennsylvannia. However studies of Icelandic, American and Irish pedigrees failed to find linkage with the chromosome 11 markers.

It may be very difficult if not impossible to establish linkage in schizophrenia not only because of the diagnostic problem but also because of the decreased reproduction by affected individuals and the rarity of large multiple affected families. There was much interest when in 1988 linkage with a high degree of statistical certainty was demonstrated between schizophrenia and two chromosome 5 probes in two English and five Icelandic pedigrees. In these families schizophrenia appeared to be inherited as an autosomal dominant condition with high penetrance, and out of 104 family members at risk and Irish, 39 developed schizophrenia. Depressingly these findings and studies in other parts of the world did not show this linkage. A genetic locus for schizophrenia without the pseudo autosomal region of the chromosome has even been proposed but not supported by any evidence so far. It is the pseudo-autosomal region in part of the distal ends of the short arms of X & Y chromosomes that undergo recombination in male meiosis.

Recently attention has been drawn to the possibility that unstable DNA sequences may be involved rather like the recent finding of the unstable tandem sequence in the Fragile X Syndrome. Of even more recent interest, is that the gene for Huntington's disease has been identified and again is characterised by an expanded tandem repeat sequence. Another similar finding was with myotonic dystrophy. These disorders have puzzling phenotype variability. As schizophrenia is another example of a syndrome that is "badly behaved" from the point of Mendelian expectations, the hunt is now on for evidence of anticipation and/or imprinting. So far this has been to no avail. The British Journal of Psychiatry, May 1994, reported that among multiple affected pedigrees age of onset among parents and offspring

were examined for evidence of both anticipation and genetic imprinting. The results did not suggest either mechanism is operating in schizophrenia but because of the small numbers only very large effects could be confidently excluded.

Overall it is likely that major mental illnesses may be only a spectrum of disorders. It may be necessary to expand the diagnostic categories to include schizophrenia, manic depressive disorder and indeed minor depression, alcoholism and even personality disorders. It would also not be surprising if a number of genes are involved. All cases of functional mental illness may not have the same underlying cause. A number of apparently discrete clinical disorders break down into a variety of molecular pathologies. One such example is retinitis pigmentosa, which is known to exist in X-linked and autosomal dominant and recessive forms. It may well turn out the vulnerability to mental illness may be a result of a biochemical abnormality in some final common pathway and that a variety of genetically induced enzyme defects can produce this.

Therefore despite some flickering promise it may be sometime before mental illnesses overwhelms the Directory of the Association of Genetic Support of Australasia AGSA).

Glossary of Terms

Allele - one member of a pair of genes at a given region of DNA.

Amyloid - A complex material composed partly of proteins which is present in the body of people suffering from several diseases.

Anticipation - the occurrence of a genetic disorder at earlier age and/or at greater severity in successive generations.

Autosomal Dominant - determined by a gene on one of the chromosomes other than the sex chromosomes, requiring only one member of the gene pair to be altered to express the condition.

Chromosome - chromosomes contain genetic material and are in every cell of the body.

Factor IX - blood coagulation factor.

G6PD - the important red-cell enzyme defect, glucose 6-phosphate dehydrogenase deficiency, is particularly prevalent in parts of the Middle East, Mediterranean, South East Asia and in people of African decent.

Heterozygous - an individual with two different alleles of a gene region.

Hypercholesterolaemia - a condition which leads to elevated levels of plasma cholesterol.

Imprinting - where gene expression is influenced by the parental origin.

Linkage - refers to genetic factors which are inherited together, because they are closely located to one another.

Meiosis - the process of cell division leading to formation of eggs and sperm with halving of the chromosome number.

Mendelian - following the patterns of inheritance proposed originally by Gregor Mendel.

Multifactorial - determined by multiple genes and also by non-genetic factors.

Phenotype - the visible expression of the action of a particular gene; the clinical picture resulting from a genetic disorder.

Polygenic -determined by multiple genes.

Tandem Repeat Sequence - a series of identical DNA sequence lying adjacent to each other in the same orientation, within a much larger DNA sequence.

MOOD DISORDERS IN CHILDREN AND YOUNG PEOPLE

Dr Meg Smith.

If your family has a number of people in it who have had serious depressive illness and manic depressive illness, then children in the family do have a higher probability of developing mood disorder than other children. This can create much anxiety and anxious watching of young children as they grow up. We don't have any sure way to know as yet which children will grow up and develop cyclothymic, depressive and manic depressive disorders. Even if we did, the real problem is how to help children who are having some difficulties now.

If you suspect a child is having mood problems there are a number of things that can be done to help. Being born with the mood disorder gene isn't a negative thing: it can also mean that one is likely to have a creative and complex way of looking at the world. Being able to control and manage mood swings means one can use the creativity more successfully.

* **Check out physical causes of the problem.** A lot of medical conditions (e.g. diabetes, hypoglycaemia, anorexia, multiple sclerosis) can cause depressive and mood disorder symptoms and so can viral illnesses like the flu and infections. a medical check up and hearing and eye tests can help to rule out other causes of mood problems and reassure everyone about what is working and what isn't.

People who have the genetic loading of mood disorder may also be more sensitive to anaesthetics, surgical procedures and medications such as cortisone. The genetic loading of mood disorder seems to mean that the neurochemicals which help the brain to cope with stress don't work as well in some people with a family history of mood disorder. Vitamin B supplements which can help the brain to manufacture the right neurochemicals to cope with stress are well worth trying. Most children will accept a Berocca B fizzy drink.

* **Emotional causes.** People with mood disorder experience emotional trauma the same way as everybody else - but they may take longer to get over it. If there has been some emotional trauma like death of a family member or separation of parents or brothers and sisters leaving home, allow the child to take a bit longer to get over it. There is no set time on grief and crying and the person will eventually recover from the experience.

Reassurance, counselling, helping the child to cope with day to day tasks and helping to talk through feelings can help the person to understand what is happening and learn to manage emotional stress in future. Emotional upset can interfere with eating and sleeping - encouraging proper eating and ensuring the child is sleeping eight hours in a twenty four hour cycle can help to establish a normal routine.

People with mood disorder may not be able to cope well with multiple emotional stresses - serious mood disorder in adulthood is often triggered by two or three emotional stresses happening close together. Because a person with mood disorder may take longer to recover from the emotional stress, events happening close together have a cumulative effect. The emotional overload then means the person cannot sort out one feeling from

another or make decisions about what has to be done next. Medication to calm down the emotional turmoil for a short period of time can sometimes be helpful.

* **Symptoms of depression.** Children show depression differently to adults. They often cannot say they are depressed or describe their feelings. Constipation, lethargy, not wanting to get out of bed in the morning and bursting into tears at slight frustration can be symptoms. Feeling there is no point in doing things, feeling friendless and that no one loves you are all common signs of low self esteem brought on by depression.

These symptoms are often overlooked in children or thought to be part of "normal growing up". Maybe the best way to treat them is as part of normal growing up in a young person who is more sensitive to the world around and his or her own feelings about it. Depression makes one unable "to see the light at the end of the tunnel" so reassurance, encouragement to just get this task done and just cope with today can help enormously. Depression also impairs complex thinking so some help may be needed to break down tasks into manageable chunks. Antidepressant medication can relieve some of the symptoms of depression so it is worth discussing possible medication with your child's doctor.

* **Biorhythms and Biocycles.** Mood disorder is really about disruption in biorhythms. So sleeping patterns, activity patterns and processing of information can be disrupted. People who are depressed often have difficulty getting up in the morning, are grumpy when they do and often aren't able to process complicated information early on. So a maths lesson first thing at school can be a horrible thing to face. They will get better as the day progresses and may do their best work in late morning and after lunch. Evening is often a good time to do creative hard work.

Work out with the young person the best times of the day for him or her and try to schedule difficult tasks at times when he or she is best able to do them. Young people who are depressed can often do well at school if class performance is less important than the homework that the person can do at a time when they function best.

Depressive cycles do pass and many adults with mood disorder accept that they have periods of low functioning but that they more than make up for it when they are not depressed. Unfortunately, rigid achievement expectations can put a lot of pressure on a person who is going through a depressive period and make the depression worse.

* THINGS YOU CAN DO TO HELP A CHILD WITH MOOD PROBLEMS

* **Start a feelings diary** - What parts of the day are better to do things in? What helps when one is feeling grumpy and can't face the work? When do I feel sleepy or happy or confident? What things make me feel bad?

* **Time out and being with others.** Everyone needs time to slow down and sort out what is going on. At times of emotional stress, quiet space can help to reduce the overload. Being with lots of people when you're feeling irritable and gloomy can be a trigger for tantrums and despair about not coping. Give the young person the choice to be alone or with someone he or she feels comfortable with and who is accepting. Don't let it be seen as punishment for not being able to cope. Tears brought on by family and social overload should not be dealt with by sending the child off to his or her

bedroom without any reassurance or comforting.

* **Medication.** If medication suggested, keep track of any symptoms and changes. Is the medication helping? Is it producing any unwanted side effects? e.g. drowsiness, weight gain or loss.

* **Keep a check on nutrition.** People who are depressed often crave carbohydrates. Carbohydrates are fine since they take a while to digest and produce the needed neurochemicals but it is the sugar and fat that often go with them that create the problems. Sugar can produce hypoglycaemic swings which make the mood swings worse. Snacks of low fat, high carbohydrate and low sugar can help to even out the blood sugar levels. People who are depressed often don't eat breakfast, crave carbohydrates by lunchtime and eat large meals at night when they are feeling better. So the person gains weight since the body doesn't digest and use up kilojoules overnight.

* DAILY NUTRITION PATTERN TO HELP WITH DEPRESSION

* **Small nutritious breakfast to stimulate metabolism,** easily digested, and attractive to a grumpy/don't want to get out of bed person: fluids such as milk, fruit smoothie, fruit juice, Vitamin B drink, stimulant such as tea, chocolate or coffee, lightly cooked egg or other protein dish, complex carbohydrate such as cereal or toast.

* **Snacks of high carbohydrate/low fat/low sugar content,** e.g. fruit, dried fruit wholemeal biscuits throughout the day to even out blood sugar levels.

* **Lunch and dinner** - same as everyone else!

Meg Smith is a psychologist who could never make it to morning lectures or to college breakfasts but did well in subjects that could be swotted up at night. If you'd like to add to this paper with your own experiences of what helped you as a child with a mood disorder please contact: Dr. Meg Smith, Department of Youth Work and Justice Studies, University of Western Sydney PO Box 555 Campbelltown NSW 2560 772 9299 or 660 7413.

CAN A FAMILY BE DEPRESSED?

Stewart L Einfeld
Associate Professor
School of Psychiatry
The University of New South Wales

Depression can be both a common and ordinary mood state or a psychiatric disorder. Manic depressive illness and Seasonal Affective Disorder are psychiatric disorders. However, in these conditions depressed mood is associated with other changes in thinking and bodily function. For example, in severe melancholic depression, depressed mood can be associated with changes in sleep, weight, motor activity and serious distortions of thinking. However, depressed mood without other symptoms is an almost universal human experience at some period of life and in it's mildest form blends in with ordinary disappointment, pessimism and unhappiness in response to ordinary life tribulations.

A major cause of sadness in a family is the discovery of a serious genetic disorder in one or more members. The human response to such a discovery can be understood as a process of bereavement which normally follows any serious loss. Such losses may be material, such as loss of a valued job, but may be more subtle such as the loss of

the family's former concept of having good health. On diagnosis, the image of good health is felt to have been lost.

You are probably familiar with the stages of bereavement process which have been widely described. That is, the first reaction is shock, often followed by denial, then anger or guilt, and then sadness, and finally some kind of resolution or adaptation. Note that sadness is one of those normal stages of response.

When can such sadness be thought of as depression? It is a matter of duration and impairment. It is hard to say how long normal sadness would last and when this is excessively prolonged. In psychologically healthy families coping with such a diagnosis there is a general trend over months or years for the sadness to diminish though it's often reawakened at particular times, when there are reminders of the original crisis time of discovering the presence of the illness.

Impairment is perhaps a better indicator of normal sadness becoming depressive disorder. If sadness is of such depth and severity that someone is unable to eat properly, sleep normally, or after a considerable time still unable to carry out their ordinary activities such as going to school or work, or if their sadness is associated with persisting suicidal ideas, then a depressive disorder has possibly developed.

Can a whole family be depressed? Diagnosis of serious genetic disorder in a family member and especially a transmissible disorder, leads to a grief response not just in one or more individuals in the family but in the family as a whole. This is because in a healthy family, each family member is to some extent influenced by the welfare of any of its members. While that bereavement process usually proceeds over time through to healthy adaptation, some families may never proceed to this stage. Where there are serious problems in family communication, allocation of family roles or unresolved legacies from the past, families can spend long periods, perhaps many years at intermediate stages of bereavement.

For example, all parents will feel guilty when anything adverse happens to their child, irrespective of whether there is any logical reason for it nor not. However, they will overcome this in time. But sometimes parents can feel deeply guilty even if irrationally so, for decades. Brothers and sisters of the sick child may also feel guilty for being healthy while their sibling is suffering, a manifestation of so called "survivor guilt". Other families find it difficult to progress through the sadness phase and cannot enjoy themselves at all, by virtue of one of the family members being disabled.

What can be done to help families avoid depression in the face of genetic disorders in one or more of its members.

Families should encourage open and frank communication about the conditions affecting them as far as possible based on realistic information. They should allocate roles in supporting each other which are appropriate for each members' capacity and these will change as children grow. Thirdly it should be remembered that each member of the family is to be cherished and valued equally. The ones with genetic disabilities should be neither scapegoated and devalued, nor become the central focus of the family's existence.

If you feel your family is having chronic difficulties in adjusting emotionally to the presence of Genetic Disorder, contact AGSA who can refer you to appropriate health professionals who are practised in assisting families with their adaptation.

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.

****

BATTEN'S DISEASE

A Tasmanian family is seeking contact. Please contact AGSA for further details.

CONGENITAL ADRENAL HYPERPLASIA

It has been requested that a seminar be held for adults with this condition later in the year. Often the adults are forgotten and it is hoped that AGSA will address this situation in 1995/96. Please notify AGSA if you are interested.

JOUBERT SYNDROME

A mother of a two month old baby girl would like contact with another family. Please phone AGSA for details.

VON HIPPEL-LINDAU SYNDROME

AGSA now knows of three families with this condition who are eager to locate more people. The incidence is 1:30,000 so if you know of anyone please contact AGSA for further details. Quite often this condition is missed as it affects different organs (eyes, brain, pancreas, kidneys) in the body and is not seen as a whole condition.

KLINEFELTER SYNDROME

Tania has a three and half year old boy with KS and she would like to set up a support group in Victoria. If you would like more information please contact AGSA or

Tania Yard
41 Jesmond Road
Croydon Vic 3136
(03) 9723 6148

There has also been a request to start up a Klinefelter Support Group in WA. Contact AGSA for details.

THE ASSOCIATION OF GENETIC SUPPORT
OF AUSTRALASIA INC.
FUNDED BY THE NSW HEALTH DEPARTMENT
REG. CHARITY No. C.C. 2702
66 Albion Street, SURRY HILLS NSW 2010
Tel: 211 1462 Office Hours:10-2 Mon-Fr

You are invited to the

LIVING GRIEF SEMINAR

Saturday 29 JULY 1995
Time: 10am to 5pm

VENUE: CHILDREN'S MEDICAL RESEARCH INSTITUTE
214 hawkesbury Road ,Westmead

Cost: $40 for members of AGSA
$60 for non-members
(Cost includes registration, morning and afternoon tea, and lunch.)

The seminar speaker will be Dr John Rogers

Dr Rogers is the Senior Medical Geneticist at the Royal Children's Hospital, Murdoch Institute. He is also a psychotherapist in priva practice who specialises in grief and life-limiting disorders. He trained in paediatrics in Melbourne, Sheffield and London. His trainir in genetics was at Johns Hopkins in Baltimore, USA.
Since 1985 John has worked with Elisabeth Kubler-Ross at her workshops and became a staff member of that organisation. He has staffe many Australian workshops.
John has wide experience in running groups for parents and individuals affected by genetic problems.

Outline of the day

Confidentiality will be a key element of this Seminar

Welcome
1. Defining the scope of the problem
Morning Tea
2. Sharing of Experiences
Lunch
3. Teaching will include Elisabeth Kubler-Ross's contribution to:-
Understanding grief and loss
The Quadrant Theory of Development
Emotions, natural and distorted
Afternoon Tea
4. Integration and preparation for home

Please detach the slip below and return to AGSA, 66 Albion Street, Surry Hills 2010.

RSVP 1 JULY 1995

Yes I will be attending; Name _____ **Telephone** _____
Address _____
Organisation (if relevent) _____

Payment Included; Yes ☐ **No** ☐

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

66 Albion Street
SURRY HILLS
NSW 2010

Tel: (02) 211 1462

****

Support and Information Officer -
Dianne Petrie
Office Hours: 10.00am - 2.00pm
Monday - Friday

****

President
Ros Smith
may be contacted on:
Tel: (047) 51 5872

Regional Contact
Judy Rands
10 Roosevelt Avenue
WAGGA WAGGA
NSW 2650
Tel: (069) 26 1560

ANNUAL SUBSCRIPTION

Individual	$15.00
Group/Organisation	$30.00

Subscription Year
1st October - 30th September

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

* lobby government bodies, both Federal and State, for appropriate funding for genetic services