THE · ASSOCIATION
OF · GENETIC · SUPPORT
OF · AUSTRALASIA

JANUARY 1994 ISSUE 11

EDITORIAL

Happy New Year.
Welcome to our first Newsletter for '94. As previously indicated, it has been necessary to alter the presentation but the basic format will remain unchanged.

The New Year also brought some good news. Our submission to the Department of Health for a non-government grant was SUCCESSFUL - we are now in a position to employ a Peer Support/ Information Officer 20 hours per week.

It is anticipated that this person will commence duties towards the end of February or early March. As you can imagine, the Committee feels somewhat elated at the prospect of offering a more personalised service to AGSA members.

With advances in medical therapies, technologies and drug regimes, AGSA is finding it increasingly difficult to keep abreast. Therefore, we would appreciate any article from families and professionals alike, which would be of interest to our readers.

As AGSA is an Australasian group it is important that all States have a say in this, your Newsletter. Please keep us advised if your group is on the move or you have an event that needs publicising.

During 1994 it is anticipated that the following new groups will be established:

- Rare Chromosomal Disorders
- Klinefelter Syndrome (47,XXY)
- Lysosomal Storage Disorders.

If you have a particular interest in any of the above, please contact the office where a register will be kept and information will be forwarded as and when appropriate.

We trust 1994 will see AGSA become more productive and we look forward to renewing old acquaintances and establishing new friendships.

With best wishes

Ros

Ros Smith

ADVANCE NOTICE

ANNUAL GENERAL MEETING

SUNDAY 13TH MARCH 1994

COMMENCING 10.00 AM

66 ALBION STREET
SURRY HILLS

Registered Charity No.C.C.27702 ISSN 1033-8624

FAMILY COUNSELLING

The following extracts are taken from an address given by Fiona Richards, Social Worker, Department of Genetics, Royal Alexandra Hospital for Children, and Executive Committee member, AGSA. This address was given at the Charcot-Marie-Tooth 1993 Annual General Meeting.

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Being part of the Genetics Department has made me more aware of not only how many serious genetic disorders there are, but of the impact of these conditions on the person affected, and on the family. I would like to talk generally about some of the common reactions to a genetic diagnosis, and the role of Social Workers and other counsellors in helping people to cope with these reactions. Coping with the often devastating effects of a genetic disorder is a long complicated process for the individual, family and at times, the health professional. Each person's reaction to the disorder is determined by his or her own personality, family and social support networks, and sometimes by the previous generations' experience of the illness. In addition, the psychosocial and cultural background of individuals can have a strong influence on the way that genetic information is received, processed and interpreted. It si important, especially for health professionals to remember that a genetic diagnosis affects the whole family.

In discussing the various emotional reactions to genetic diagnosis, it is possible to see a parallel with the stages of emotional reaction often experienced by people facing a terminal illness and their families which have been described by Elizabeth Kubler-Ross. these reactions are denial, anger, bargaining, depression and acceptance and most people don't progress neatly from one stage to the next but rather drift in and out of the stages as time goes on.

REACTIONS UNDER SLIGHTLY DIFFERENT HEADINGS. BEGINNING WITH:

Grief: - which is the overall reaction to losses, present and future resulting from the genetic disorder. These losses may be experienced by the parents as the loss of the dream of a perfect child and by the individual. For example, loss of independence, ability to earn a living, loss of control or effective functioning of parts of the body, loss of confidence, motivation, dignity, and self-esteem, sometimes loss of friends. This grief is shared by all family members to different degrees.

Anger and Frustration: - about these losses and the difficulties experienced in day-to-day life. Some people express this anger and frustration through aggressive behaviour, others become depressed and withdrawn. Both these reactions tend to lead to further isolation.

Guilt: - is often felt strongly about the possibility or knowledge that the abnormal gene has been passed on to the children even if unknowingly. The guilt is sometimes associated with anger that someone in the family knew about the disorder but had not said anything. Associated with the guilt also can be feelings of low self-esteem, shame, worthlessness and inferiority.

Denial: - is often a factor underlying behaviour. It is a form psychological defense where a person refuses to recognise a traumatic situation. Most of

practise denial to a certain extent in our everyday lives. In some families, denial can be recognised as a refusal to acknowledge problems associated with the genetic condition. Often years of family silence and non-communication have reinforced the denial and this may be one reason for misdiagnosis. Denial operates at different levels. It may be conscious and can be dissolved if the person receives an empathic response. Or it may be operating at a deeply unconscious level defending against depressive or aggressive impulses.

The consequences of denial for families include:

1) Ignorance of the existence of the genetic disorder in the family so that decisions are made without the knowledge of genetic risks.
2) Even if genetic risks have been explained, denial can lead to having children as a way of reinforcing the belief that there is nothing to worry about.
3) Avoidance - of medical and social support services as contact with these services means facing the distressing problems that some families would prefer to ignore. Sometimes the health services are perceived as the source of the problem and so are avoided. Of course, there may be other reasons why services are avoided.
4) Distortion of Genetic Information. - It has been known that families have been told incorrect information by health professionals but even when given the correct facts people sometimes choose to believe otherwise; for example, that the disorder can skip generations, that it only affects males or females, that children who look like the affected parent will be the ones to get the disorder and so on. There is no simple solution for overcoming denial. Often it decreases with time if there is adequate support and understanding and the family gradually comes to an acceptance of their situation.

SOCIAL EFFECTS OF GENETIC DISORDERS

Social Isolation: - can result from community ignorance of genetic disorders, feelings of embarrassment or stigmatisation. Community attitude is improving, however, and there is more acceptance of disabilities than there was 10 or 20 years ago. Self-help associations such as yours play a very important role in promoting public awareness of genetic disorders and encouraging more understanding.

Stress: -associated with caring for someone with a disability, restriction of social activities, financial difficulties, anxiety about genetic implications and the future of children. This can put a strain on relationships both within and outside the family, for example, marriage and work relationships.

Effects on Children: - of growing up with a parent who has a genetic condition. These will vary according to the way in which the parent is affected and how this parent and the unaffected parent are seen to be coping. Often members of the extended family can be an additional source of comfort and support for children for example, grandparents, aunts and uncles, and cousins.

Children can react to anxiety and stress within the family by experiencing behavioural problems. These will usually decrease when their fears and concerns have been addressed.

BENEFITS OF COUNSELLING

These fall into two areas - information and support. A Social Worker (or other counsellor) aims to facilitate the best possible adjustment to the genetic diagnosis over time for the individual or family. During the initial phase of shock, disbelief and grief, the Social Worker can be supportive and encourage the family's expression of feelings reassuring them that these are normal reactions. At appropriate times, the Social Worker can repeat and clarify information the family has been given about the condition. As time goes on, the Social Worker can offer guidance on how best to explain to other relatives and friends about the condition, which should help to minimise any communication problems with the family's social network.

The Social Worker can also act as an advocate for the family, if necessary, in dealing with the health system. Social Workers can provide information bout resources available in the community including financial benefits.

Social work contact with families affected by a genetic disorder may be brief or ongoing, depending on the severity of the condition and the family's reaction.

Families and affected children may experience crises at significant stages of development; for example starting kindergarten or school and during adolescence. At these stages parent and child are confronted once again with the reality of the condition and differences from others. This may precipitate an emotional crisis for those who have not come to terms with, or accepted the reality of the condition. Social work counselling and possibly even referral for psychiatric assessment or family therapy can be very beneficial in these situations.

People affected by a genetic condition feel initially that they are the only ones in their situation. Meetings with others through Associations such as yours, who have been through the experience and coped with the various crisis times, can be invaluable. Support groups offer the opportunity to express feelings openly with accepting and understanding peers. In my experience, support groups are also the best source of up-to-date, accurate and understandable information about the genetic disorder.

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RESOURCES AND AIDS

CATHY HOUSE

The Lions Club of Tin Can Bay Inc., recently wrote to AGSA. The correspondence included brochures and details of **Cathy House**, a holiday camp on the foreshores of the Great Sandy Straits.

The house caters for up to 44 people and has a huge entertainment dining room, large commercial kitchen, laundry and inground pool with ramp access for the disabled. The bedrooms are 4 to 6 birth and the cost is $6.00 per head per day.

Day trips to Cooloola National Park, Fraser Island and the coloured sands can be arranged.

For further bookings or enquires contact John Hunter, (074) 86 4269, or write to the Lions Club of Tin Can Bay Inc, c/- Post Office, Tin Can Bay, Qld 4580.

PROFILE A - Z GENETIC CONDITIONS

It is the intention of AGSA to profile each Support Group/Disorder alphabetically 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.

For your information we profile.....

ANDERSON-FABRY DISEASE

Reprinted from the Information Sheet prepared by Margaret Davie

The Disease

This is a rare inherited metabolic disorder which is progressive and, in affected men is the cause of shortened life-span.

Anderson-Fabry disease is passed on from generation to generation. Carrier women have no (or very mild) symptoms, but affected men show the full range of symptoms characteristic of the disease.

Metabolic diseases occur when there is a metabolic block because a catalyst or enzyme necessary to perform essential chemical reactions in the body, is absent or malfunctioning.

In Anderson-Fabry disease, this enzyme is alpha galactosidase A. This defect results in the slow build-up of a substance, glycosylated ceramide, in the body.

Symptoms

Characteristic symptoms in affected men are shooting pains in the arms and legs, which often develop in childhood. There may also be unexplained episodes of raised temperature and stomach pains.

Diagnosis is rarely made before early teens, when a characteristic purplish rash bottom, navel or around the genital area. Slight bleeding or spotting may occur from the rash. The disease is characterised by attacks of pain and raised temperature, which come and go. Later complications include cataracts, kidney failure, stroke, heart murmur and rarely, heart beat irregularity. If a renal transplant needs to be carried out, the transplanted kidneys are not affected with Anderson-Fabry disease. Carrier females may occasionally suffer from pains but other symptoms are very rare.

Genetics

The faulty gene for Anderson-Fabry disease is coded on the X chromosome. As females have two X chromosomes, a mistake on one X will not cause the disease, but they will be carriers. Their chance of having an affected son or a carrier daughter is 1 in 2 or 50/50.

Males have only one X chromosome and a mistake will produce the disease. All their daughters will be carriers, but all their sons will be free from the disease. In the great majority of families this mistake on the X chromosome is new, starting with the carrier mother or the affected son.

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CONTACT CORNER

AGSA will publish requests for contact 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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Sturge-Weber Syndrome

Sue Clough mother of a 5 year old son with Sturge-Weber Syndrome, would welcome contact from any other family. Sue can be reached on (02) 363 5036 or by post at 45 Jersey Road, Woollahra, NSW 2025.

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A family of a 4 year old child with MOSAIC TRISOMY 13 would like to have contact with a family with similar experiences.

If you can be of assistance please contact
Ms Annette Henry
Genetic Counsellor
The Orana Community Health Services
P.O. Box 739
DUBBO NSW 2830
Tel: (068) 85 8999

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Hartnup Syndrome

AGSA is seeking contact for one of our members. Please telephone the office if you can be of assistance.

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The mother of an eight and a half year old daughter with a deletion of Chromosome 18q22.1 seeks contact with anyone who has a child similarly affected. Please telephone the AGSA office if you can be of assistance.

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Mother of a daughter with **Sotos Syndrome** wishing contact. Further particulars from the AGSA office.

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SUPPORT GROUPS

AUSTRALIA

ANDERSON-FABRY DISEASE

29 Pope Ave.,
BORONIA VIC 3155
Tel: (03) 762 3910

Dear Sir/Madam.

I am interested in setting up a Support Group for families and sufferers of Anderson Fabry's disease and I would appreciate if the attached information sheet regarding this disease is displayed throughout your facilities.

To my knowledge there is no Support Group in Australia for Anderson Fabry disease, (there is little known about it) and it is my intention to form a Group to raise funds and awareness for research into this disease.

Hoping you can assist me.

Yours sincerely,

Margaret F. Davie

INTERNATIONAL

NEW GROUPS

Apert Syndrome
Ann Luxton and Howard Esler
6 Peter Mulgrew Street
Avondale
AUCKLAND 1007 NZ
Tel: (09) 627 9137

Craniofacial Support Group
A number of interested parents will be working to develop a support group for craniofacial conditions. Among these conditions are Craniosynostosis, Craniostenosis, Aperts, Crouzon, Saethre-Chotzen and Sagittal Synostosis. More information can be obtained by contacting

Linda Partridge,
Development Officer
Contact a Family
16 Strutton Ground
London SW1P 2HP.

ASSERT - Angelman Syndrome Support Education and Research Trust
Maybe Cottage
41 George Lane
Marlborough
Wilts SN8 4BX
Tel: (0672) 515057

GROUPS ON THE MOVE

Coloboma Support Group
43 Dinam Road
Caergeiliog
Holyhead
Gwynedd LL65 3ND
Tel: (0407) 741413

Cri du Chat Syndrome Support Group
49 Summerside Place
Edinburgh EH6 4NY
Tel: (031) 554 2524

Ehlers Danlos Support Group
1 Chandler Close
Richmond
North Yorks DL10 5QQ
Tel: (0748) 823867

Tourette Syndrome (UK) Association
Valley Mead
27 Monkton Street, Ryde
Isle of Wight PO33 2BY
Tel: (0983) 568866

From Alliance - USA :

UPCOMING MEETINGS

Annual Clinical Care Symposium of the National Neurofibromatosis Foundation - March 13; Hyatt Orlando, Kissimmee, Florida. Contact Fran Morris, National Neurofibromatosis Foundation, 141 Fifth Avenue, Suite 7-S, New York, NY 10010; Tel: 212/460-8980.

First Meeting of the American College of Medical Genetics - March 15-17; Hyatt Orlando, Kissimmee, Florida. Call 301/571-1825.

Children and Hospitals Week - March 20-26. For information on how to get involved, contact Trish McClean, ACCH, 7910 Woodmont Ave., Suite 300, Bethesda, MD 20814; Tel: 301/654-6549.

National Tay-Sachs & Allied Diseases Association National Conference - April 22-24; Toronto. Contact Debi Gutter, NTSAD, 2001 Beacon St., Brookline, MA 02146; Tel: 617/277-4463.

Sjogren's Syndrome Foundation Symposium - April 23; NIH, Bethesda, MD. Contact the Foundation, 382 Main Street, Port Washington, NY 11050; Tel: 516/767-7156.

American Cleft Palate-Cranio-facial Association Annual Meeting - May 17-21; Westin Harbour Castle, Toronto. Contact ACPA, 1218 Grandview Ave., Pittsburgh, PA 15211; Tel:412/481-1376.

Association for the Care of Children's Health Annual Conference - May 22-25; Westin Harbour Castle, Toronto. Contact Donna-Renee Price, ACCH, 7910 Woodmont Ave., Suite 300, Bethesda, MD 20814; Tel: 301/654-6549. Parents are urged to apply early for travel and hotel stipends to attend the parent part of the conference.

Spina Bifida Association of America Annual Conference - June 22-25; Radisson Hotel, Keystone Crossing, Indianapolis, IN. Contact Heidi Bulow, SBAA, 4590 MacArthur Blvd., Suite 250, Washington, DC 20007; Tel: 202/944-3285.

Cornelia de Lange Syndrome Foundation 15th Annual Convention - June 23-26; Radisson Tempe Mission Palms Hotel, Tempe, AZ. Contact CdLS Foundation, 60 Dyer Ave., Collinsville, CT 06022; Tel: 1/800/233-8355.

National Organization for Albinism and Hypopigmentation International Conference - July 14-17; Adam's Mark Hotel, Philadelphia, PA. Contact NOAH, 1500 Locust St., Suite 1816, Philadelphia, PA 19102; Tel: 1/800/473-2310 or Br. William Harkin at 215/446-0202.

Williams Syndrome Professional Conference - July 27-28; Contact Nancy Grejtak, WSA, 605 Giralda Drive, Los Altos, CA 94024; Tel: 415/948-7604. **National Convention** - July 28-31; San Diego, CA. Contact Sally Meersman, WSA, 2841 Highridge Rd., La Crescenta, CA 91214; Tel: 818/249-4261.

AGSA
66 Albion Street
SURRY HILLS
NSW 2010
Tel: (02) 211 1462

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

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

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SUBSCRIPTIONS

Individuals - $15.00

Groups/Organisations - $30.00

Subscription Year
1st October - 30th September

AGSA aims to:

* 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

* provide a contact point for families who are affectd by genetic conditions so rare that they do not have their own support group

* facilitate accessability 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