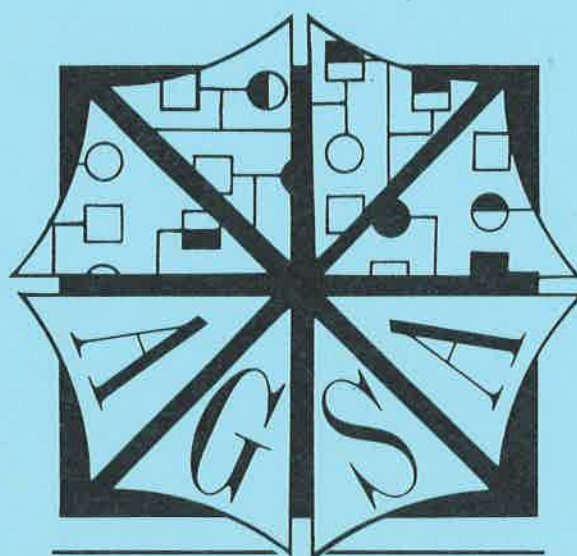


A G S A

NEWSLETTER

WINTER 1992

ISSUE 9



THE • ASSOCIATION
OF • GENETIC • SUPPORT
OF • AUSTRALASIA

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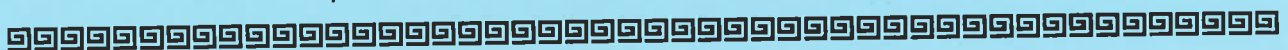


SUBSCRIPTIONS

Individuals — \$15.00

Groups/Organisations — \$35.00

Subscription Year — 1st October to 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 affected by genetic conditions so rare that they do not have their own support group
- facilitate accessibility 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.

* * * * *

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

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- EXECUTIVE DIRECTOR'S REPORT
- GENETIC SERVICES
- SUPPORT GROUP NEWS BULLETIN
- A FAMILY STORY
- CONTACT CORNER
- SEMINARS/WORKSHOPS
- RESOURCES AND AIDS
- PROFILE — A-Z GENETIC CONDITIONS
(Insert — *Please remove if required for your files*)
 1. Ectodermal Dysplasia
 2. Sanfilippo Disease

IN FUTURE ISSUES ...

- Glossary of genetic terms
- Commonwealth/State Disability Agreement — implications for families.

PRESIDENT'S REPORT

Whilst this newsletter is overdue, we thought it important that the first newsletter produced by the new committee should maintain the trend established by Ros Smith in quality and quantity.

I am sure you will find this newsletter informative and I wish to particularly compliment our Executive Director, Mary Ford, for her contribution.

At the Annual General Meeting in March, I stated that AGSA was very much at the crossroads and its survival in the 1992/93 year would very much depend on the commitment of not only the committee and its members but also the ability to raise sufficient funds to ensure the future viability of the association.

As a first step Mary Ford has been in constant contact with member support groups and in the main the reaction has been very positive. This is covered in Mary's report.

Your committee is concentrating on an awareness campaign to increase our exposure throughout the community. The steps we have instigated include:

1. The production of an informative new brochure which will be available to the public, medical and health professionals, and support groups. A copy is enclosed in this newsletter.
2. We have retained a public relations consultant who is presently holding discussions with two major consumer magazines which will hopefully result in the publication of feature articles over the next two or three months.
3. A radio commercial has been recorded and copies have already been requested by radio stations 2UE in Sydney and 2AD in Armidale. We propose issuing copies to stations throughout the State to coincide with our fund raising campaigns.

We have instituted a new fund raising campaign which entails the sale of low cost products. Although only in its infancy the campaign is already proving successful and we believe it will underwrite our fixed costs in this financial year. In addition our public relations consultant has been asked to consider the following initiatives which we expect will ensure the long term financial viability of AGSA. These are not short term programs but more attuned to the next three to five years. They are:-

1. Sponsorship for newsletters, seminars, brochures etc.
2. Direct grants from public companies or individuals for specific programs.
3. An Art Union.
4. Direct mail campaigns.

We are confident that these initiatives will ensure that AGSA not only achieves a higher profile in the community but also financial stability which will enable it to fulfil its objectives and be of benefit to all those who it represents.

R. J. MacDiarmid
President

EXECUTIVE DIRECTOR'S REPORT

As this is my first report in AGSA's Newsletter it is quite lengthy but I feel it is important every one is aware of AGSA's activities. I would also like to take this opportunity to say I am excited about my appointment as AGSA's Executive Director and also about the activities which AGSA is proposing.

I believe AGSA has a significant role to play in assisting those affected by an inherited disorder and by improving networking of Support Groups and Medical and Allied Health Professionals throughout Australia.

COMBINED SUPPORT GROUPS MEETING REPORT

On Monday 27th April, 1992 a combined meeting of Member Support Groups was held.

This meeting was attended by representatives of AGSA and individual Support Groups.

The aims of the meeting were to:

- * bring Support Groups up to date with AGSA's activities
- * see how Support Groups perceive AGSA's role
- * discuss proposed activities

a) AGSA'S BACKGROUND INFORMATION

Over the years Support Group committees change therefore an overview of why AGSA was formed was presented.

This included:

- Rationale for forming AGSA
- AGSA's aims - plus addition to aims
- Achievements
- Future Plans
- AGSA's requirements

b) MARKETING CONSULTANT

A Marketing Consultant, Mr Stuart Hudson, has been employed by AGSA two days per week for a period of six months. This will ensure awareness regarding AGSA's activities is carried out in the most effective manner and in conjunction with fundraising activities being organised.

c) SUGGESTIONS HOW AGSA CAN ASSIST SUPPORT GROUPS

Short Term

- * Use of AGSA's Equipment (eg photocopier)
- * Insurance policy as an umbrella group
- * Emotional Support
- * Seminars
- * Condition Management
- * Information Referral point - resources available
- * Kits - How to Start a Support Group
- * Ensure Support Groups in isolated areas receive information from seminars held

Long Term

- * Financial Assistance given when a new Support Group is being formed
- * Lobby Government
 - obtain grants
 - assistance with pharmaceutical payments
- * Accommodation when families from isolated areas visit metropolitan area for medical treatment
- * AGSA to meet with Support Groups, from isolated areas, in a central location, at least once a year
- * Respite Care

d) SUGGESTED SEMINARS

- * Support Group Information Day
 - how to become Incorporated
 - how to gain sales tax exemption
 - how to gain tax exemption
 - how to produce a newsletter
 - suggestions for fundraising activities
- * General Counselling Skills
- * Care for the Carer
- * Education for Peer Support — Do's and Don'ts of Peer Support
- * How genetic disorders in a family affect family dynamics
- * Combined Medical and Allied Health Professionals together with those affected by a Genetic Disorder
- * Recent research technological developments

At the meeting it was felt there is an urgent need for a Seminar/Workshop to be conducted on General Counselling Skills/Leadership Skills. This is to be conducted on 23rd August, 1992. Full details are included in this Newsletter.

e) REGULAR MEETINGS WITH SUPPORT GROUPS AND AGSA REPRESENTATIVES

An informal meeting be held on a six monthly basis between Support Groups and AGSA representatives.

f) OUTCOME OF MEETING

Information about what was discussed at this meeting, the outcome, together with a questionnaire, were forwarded to all member Support Groups throughout Australia.

GENERAL INFORMATION

1. New Brochure

A new brochure outlining AGSA's activities, member Support Groups etc has now been printed and will be used in conjunction with fundraising activities; forwarded to Medical and Allied Health Professionals; Community Health Centres etc.

2. Awareness Programme

We are directing our endeavours toward:

- * Women's Magazines
- * Australian Dr (and similar publications)
- * Local and regional newspapers
- * Local and country Radio Stations.

3. Invitation to Non-Member Groups

As many member Support Groups have indicated they are interested in meeting with other Support Groups who experience similar problems, I have written to Non-Member Support Groups to ask them if they would like to be involved. This would enable the best networking of support to take place.

4. Resources Available in Each State

It is imperative that information on resources available throughout Australia is collated. For this reason I have contacted other States requesting a list of resources which are available at hospitals for individuals and families as well as all Genetic Support Groups which have been set up in each State. If anyone has information on these two items I would be grateful to receive it.

5. Visits to Individual Support Groups

I am very much aware of the time constraints on individuals and families affected by a genetic condition. Due to this I have been attending meetings with individual Support Groups which I feel has been very worthwhile. If you would like me to attend one of your meetings please let me know as I feel it is the best way for me to understand problems which Groups may be experiencing and to discuss how AGSA can assist.

If you would like to discuss any matters with me or feel I can assist in any way please don't hesitate to contact me on (02) 416 0647.

I look forward to meeting you.

Until next time, take care.

Mary Ford
Executive Director

GENETIC SERVICES

WHO IS THE GENETIC COUNSELLOR?

HOW CAN HE/SHE BE OF HELP TO ME?

The genetic counsellor is a health professional who is trained in the areas of genetics and counselling.

The role of genetic counsellors varies according to the needs of their particular units. Some are situated in metropolitan areas and others more autonomously in country areas (outreach) but allied to the metropolitan service.

Genetic counsellors are usually the first point of contact for families with genetic referrals and are often responsible for co-ordinating appointments. In addition, they may act as a resource for education information about a diagnosis, as a link to support groups and a patient care co-ordinator through allied professional services.

Addressing emotional issues at the time of consultation is an integral and important aspect of overall patient care. Genetic counsellors understand the trauma and distress which may accompany a genetic diagnosis and the need to talk about fears and anxieties which arise.

You may wish to have someone talk to your child's school or pre-school teacher or class-mates. Perhaps talking to other family members is not easy. You may be worried about how they will respond, or anxious if you think they will be upset.

AGSA members may be interested in the following updated list of names and addresses of Genetic Counsellors employed in Australia.

NEW SOUTH WALES

Camperdown

Ms Bronwyn Butler (02) 692 6273
Genetic Counsellor
Ms Mona Saleh (02) 692 6142
Genetic Counsellor
Medical Genetics and Dysmorphology Unit
Royal Alexandra Hospital for Children
PO Box 34
CAMPERDOWN NSW 2050
Ms Fiona Richards (02) 692 6273
Genetic Social Worker
HD Predictive Testing Programme
Royal Alexandra Hospital for Children
PO Box 34
CAMPERDOWN NSW 2050

North Shore

Ms Miriam Kaganer-Briet (02) 438 7280
Genetic Counsellor, MSAFP Program
Ms Lynn Rawlings (02) 438 7280
Genetic Counsellor, MSAFP Program
C/- Ground Floor, Maternity
Royal North Shore Hospital
ST LEONARDS NSW 2065
(Fax: (02) 906 1872)

Paddington

Ms Gillian Brown (02) 339 4432
Genetic Counsellor
Royal Hospital for Women
188 Oxford Street
PADDINGTON NSW 2021

Randwick

Ms Robyn Pedersen (02) 399 2156
Genetic Counsellor
Dr Samantha Wake (02) 399 2950
Genetic Counsellor
Department of Medical Genetics
Hut J
Prince of Wales Children's Hospital
High Street
RANDWICK NSW 2031
Ms Hazel Robinson (02) 399 2294
Co-ordinator, Fragile X Program
Hospital School
Prince of Wales Children's Hospital
High Street
RANDWICK NSW 2031

Westmead

Ms Meryl Smith (02) 633 6892
Genetic Counsellor
Department of Paediatrics
Westmead Hospital
WESTMEAD NSW 2145

Newcastle

Ms Jan Roberts (049) 60 2206
Genetic Counsellor
Regional Medical Genetics Unit
Newcastle Western Suburbs Hospital
PO Box 21
WARATAH NSW 2298
Ms Ann Colhoun (049) 21 3600
Co-ordinator, Prenatal Diagnostic Unit
John Hunter Hospital
Locked Bag 1, Newcastle Mail Centre
NEWCASTLE NSW 2301
(Fax: (049) 21 3999)
Ms Sue Latham (049) 28 0940
Genetic Associate
Stockton Centre
STOCKTON NSW 2295

NSW COUNTRY OUTREACH SERVICES by Region

Central Coast

Ms Lucy Cook (043) 23 3166
Genetic Counsellor
Public Health Unit
PO Box 172W
WEST GOSFORD NSW 2250
(Fax: (043) 23 6276)

Central Western

Ms Kim Frumar (063) 33 1311
Genetic Counsellor
Ms Gillian Shannon (063) 33 1311
Genetic Counsellor
C/- Bathurst Base Hospital
Howick Street
BATHURST NSW 2795

Far West

Ms Glenys Warren (080) 88 5800
Genetic Counsellor
Community Health Centre
BROKEN HILL NSW 2880

Hunter

Ms Jacqueline Gaul (065) 45 1720
Genetics Clinic Co-ordinator
Musswellbrook Community Health Centre
MUSWELLBROOK NSW 2332

Illawarra

Ms Judith Elber (042) 28 4177
Genetic Counsellor
Illawarra Area Health Service
Illawarra Child Development Centre
Porter Street
NORTH WOLLONGONG NSW 2500

New England

Mr John Rae (067) 66 2555
Genetic Counsellor
PO Box 83
TAMWORTH NSW 2340
(Fax: (067) 66 6638)

North Coast

Ms Lucille Stace (066) 20 2493
Genetic Counsellor
Lismore Base Hospital
PO Box 419
LISMORE NSW 2480
Mr Chris Johnson (065) 83 3944
Genetic Counsellor
Hastings Community Health Centre
Morton Street
PORT MACQUARIE NSW 2444
Ms Dorothy Wilson (066) 52 2000
Genetic Counsellor
Coffs Harbour Community Health Centre
38 Gordon Street
COFFS HARBOUR NSW 2450

South Eastern

Caroline Willis (048) 21 6244
Genetic Counsellor
Child Development Unit
Goldsmith Street
GOULBURN NSW 2580
(Fax: (048) 21 9660)

South Western

Bernadette Taylor (069) 21 5755 Ext 393
Genetic Counsellor
Wagga Wagga Base Hospital
PO Box 159
WAGGA WAGGA NSW 2650
(Fax: (069) 21 5632)

SOUTH AUSTRALIA

Sister Sue White (08) 267 7374
Co-ordinator
Ms Elizabeth Burton
Genetic Counsellor
Medical Genetics Unit
Adelaide Children's Hospital
NORTH ADELAIDE SA 5006
Ms Clare Said
Obstetrics and Gynaecology
Queen Victoria Hospital
160 Fullerton Road
ROSE PARK SA 5067

VICTORIA

(Outreaches to Tasmania)

Ms Margaret Olsen (03) 345 5045
Genetic Counsellor (03) 345 5157
VCGS
Murdoch Institute
Royal Children's Hospital
Flemington Road
PARKVILLE VIC 3052
Ms Ann Robertson
Genetic Counsellor
Royal Womens Hospital
Gratten Street
CARLTON VIC 3053

Ms Mary-Ann Young C/- (03) 345 5157
Genetic Counsellor (03) 345 5158
Monash Medical Centre
Locked Bag 29
CLAYTON VIC 3168

WESTERN AUSTRALIA

Ms Jill Bain
Senior Social Worker
Genetic Services, Agnes Walsh House
King Edward Memorial Hospital for Women
Bagot Road
SUBIACO WA 6008

SUPPORT GROUP

NEWS BULLETIN

Your chance to publicise and provide information about your Society's or organisation's upcoming events and new developments.

* * * * *

The following is a list (of **AGSA Member Support Groups**) for your information:

Albino Support Group
Alliance (USA)
Als-Motor Neurone Disease Association Inc
Batten Disease Support Group (ACT)
Care (Scotland)
Charcot-Marie-Tooth Association Aust
Charcot-Marie-Tooth Disease Muscular Atrophy Int (Canada)
Cleft Palate and Lip Society
Coeliac Society of NSW Inc
Contact A Family (UK)
Cornelia de Lange Syndrome
Cystic Fibrosis Association of Queensland Limited
Cystic Fibrosis Association ACT
Cystic Fibrosis Association VIC
Cystic Fibrosis Foundation NSW
D.E.B.R.A.
Depressive and Manic Depressive Association of NSW
Aust Foundation for Ectodermal Dysplasia
Ehlers-Danlos Syndrome Support Group
Fragile X Support Group
Friedreich Ataxia Association of NSW
Friends Incorporated (QLD)
Friends in VIC Inc
Genetic Interest Group (UK)
Genetic Metabolic Disorders Support Group (VIC)
Haemophilia Society of NSW
Handital
Hunter Valley Genetic Support Group
Huntington's Disease Aust (NSW) Inc
Huntington's Disease Aust (QLD) Inc
Huntington's Disease Wellington (NZ)
Leukodystrophy Foundation (USA)

Little People's Association
 Little People's Association of Australia (VIC)
 Lowe's Syndrome Association Inc (USA)
 Marfan Syndrome Support Group (VIC)
 Marfan Syndrome Support Group
 MPS Diseases and Related Disorders Society
 Muscular Dystrophy Association of NSW
 Muscular Dystrophy Association (NZ)
 Neurofibromatosis Association of Australia
 Noonan Syndrome Support Group
 Osteogenesis Imperfecta Society of NSW
 Parent to Parent (NZ)
 Prader-Willi Syndrome Association of NSW (Aust)
 Inc
 Retinitis Pigmentosa Society NSW
 Rett Syndrome Association of Aust (VIC)
 RTMDC (UK)
 Schizophrenia Fellowship (NZ)
 Schizophrenia Fellowship of NSW
 Spina Bifida Group NSW
 Spina Bifida Association WA Inc
 Spinal Muscular Atrophy Support Group (QLD)
 Thalassaemia Group
 Thalassaemia Society of NSW
 Tuberous Sclerosis Society in Aust
 Turners Syndrome Ass Inc NSW
 Williams Syndrome Association
 More Groups are joining as they become aware of
 AGSA's activities.

* * * * *

NEW SUPPORT GROUPS

THE KLIPPEL-FEIL SUPPORT GROUP

The purpose of the Klippel-Feil support group is to provide a forum of communication, education and to seek out the best medical support where necessary. So very few people are affected by Klippel-Feil that sufferers can feel disheartened when they attempt to discuss it with others. You could assist the group to achieve its goals by simply keeping a line of communication open; your experiences could in some small way provide support to another.

Klippel-Feil simply refers to fusion of two or more vertebrae, which often restricts movement or alignment of the spine. This may cause neck or back pain or present special complications when playing sports. Some people have additional conditions that can affect either their hearing, ears, face, speech, larynx, mouth, thyroid, eyes, heart, genitourinary system, scapula, development or flexibility of the arms, hands or legs. The incidence of the Klippel-Feil syndrome has been estimated to be 2-3 in 100,000 people.

People with Klippel-Feil do not usually have other affected members of the family. Many of those with Klippel-Feil lead normal and productive lives. It is

generally believed that most cases of Klippel-Feil are not genetically inherited but the result of a disturbance to the normal pattern of development within the early embryo. A disturbance to the early development of the embryo could result from any number of possible causes, which have remained undefined in the case of Klippel-Feil. Some people with Klippel-Feil have required surgery to stabilise the spine but never to remove the vertebral fusion(s). Some reports have indicated that sufferers are often very difficult to intubate during general anaesthesia. Associated conditions, such as hearing loss, can present other challenges in association with Klippel-Feil.

For subscription to the Klippel-Feil Support Group's newsletter and social calendar please contact the secretary of the Klippel-Feil Support Group by phone:

Contact

Gail Ryan

31 Florida Crescent

Riverwood NSW 2210

Phone: (02) 53 9657

* * * * *

Yvonne and Paul Higgins

A Personal Invitation

to the first public meeting

ADRENOLEUKODYSTROPHY (X-Linked)

Tuesday, 18th August 1992

at

The Doreen Dew Lecture Theatre,
The Royal Alexandra Hospital for Children
Camperdown Sydney 2050

2.30pm

You are cordially invited to a First Public Meeting of the Adrenoleukodystrophy/Adrenomyeloneuropathy Support Group in Australia. Its objective is to share information gained on the caring of persons afflicted with Adrenoleukodystrophy/Adrenomyeloneuropathy and to offer other families in similar situations the support they need. Support Families now exist in Albany, Alice Springs, Hobart, Melbourne, Perth and Sydney.

Agenda for the Meeting will be to form a Working Party for the Support Group.

Dr Hugo Moser, M.D., University Professor of Neurology and Paediatrics, Johns Hopkins University, Baltimore Maryland, USA, an authority on:

- * Adrenoleukodystrophy (X-linked)
- * Adrenomyeloneuropathy
- * Neonatal Adrenoleukodystrophy
- * Rett Syndrome
- * Zellweger Syndrome

will be speaking on new information about Adrenoleukodystrophy and information about natural history and therapeutic interventions of Adrenoleukodystrophy at this Meeting.

Hope you can come!

R.S.V.P. Ms Mary Ford, Executive Director

The Association of Genetic Support of Australasia Inc

(02) 416 0647

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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 is hoped 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 . . .

ECTODERMAL DYSPLASIA

*Extracted from the publications of the
National Foundation for Ectodermal Dysplasias, USA*

AND

SANFILIPPO DISEASE

*Extracted from the booklet "Sanfilippo Disease"
published by the MPS Society, Buckinghamshire, UK*

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ECTODERMAL DYSPLASIA

INTRODUCTION

Ectodermal Dysplasia (ED) is not a single disorder, but a group of closely related conditions. The Ectodermal Dysplasias are heritable conditions in which there are abnormalities of two or more ectodermal structures such as the hair, teeth, nails, sweat glands and other parts of the body.

WHAT IS AN ECTODERMAL STRUCTURE?

Before the developing baby is large enough to be seen, a layer of cells covers the outside of the body. This surface layer of cells is called the ectoderm, and from it develop the skin, hair, nails, teeth, nerve cells, sweat glands, parts of the eye and ear and parts of some other organs (see Figure 1). Each of the listed parts of the body, then, is called an ectodermal structure. There are many disorders that involve only one of these structures and are not properly called ED. Any combination of defects involving more than one of these structures, however, should be called ED; the list of such combinations is extensive. For example, one person may have missing teeth and defective nails, while another may have missing teeth, inability to sweat and sparse hair. Still another may have sparse hair and a hearing loss. Each combination of defects represents another type of ED and has a specific name.

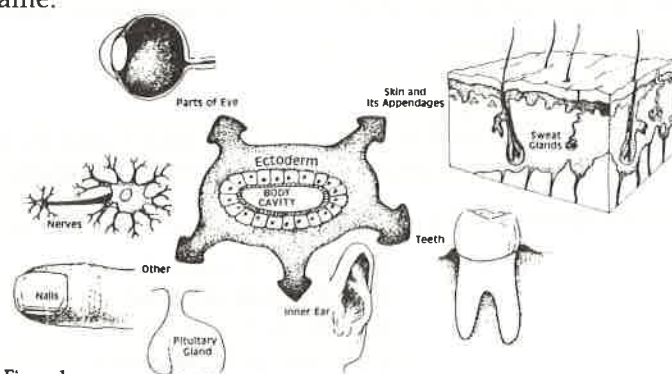


Figure 1

WHAT CAUSES ECTODERMAL DYSPLASIA?

Ectodermal Dysplasia is caused by a single abnormal gene or pair of abnormal genes. The chance for parents to have an affected child depends on the type of ED that exists in the family. In some families the mistake in the gene was a fresh mistake (mutation) in their child and the likelihood of another child being affected is very low. There are other families however that have an inherited form of ED with recurrence risk ranging from 25% to 50%.

SWEAT FUNCTION

Diminished or absent sweating is a common problem. The sweat glands are absent, reduced in number, or may not function normally. Reduced sweating may result in very high fevers, because the body regulates its temperature by sweating. Often, the first clue that the sweat glands are absent or are not functioning normally is an elevated temperature.

Elevations in body temperature are often caused by high environmental temperatures, excessive activity, or heavy clothing. When the body temperature is elevated, the skin feels dry, hot and may be flushed or pale.

HAIR

The scalp hair is absent, sparse, fine, lightly pigmented, or abnormal in texture. The hair may also be fragile and unruly, sticking out in all directions and difficult to comb. The hair is dry because the oil glands are absent or poorly developed.

Some defects of the hair are evident at birth, while others are not noted until later in life. Hair growth is slow and haircuts are not often needed. After puberty hair growth improves in some persons.

The eyebrows, eyelashes, and other body hair may also be absent or sparse, but beard growth in males is usually normal.

NAILS

Most people with ED do not have nail abnormalities, although the nails are frequently dry and rough. A distinctive finding in one of the forms of ED is a short nail that fails to grow to the end of the finger. In others, the nails may be thin and fragile, thick and distorted, or brittle and slow-growing. Nails with any of the listed abnormalities may be prone to infection.

TEETH

The teeth are missing altogether or reduced in number. Teeth that are present are widely spaced, tapered, or malformed. In persons with some types of ED, the enamel (outer layer of the teeth) is defective and there may be an excessive number of cavities. When teeth are missing the jawbones in which they are ordinarily embedded do not develop well, leading to a typical aged appearance of the face.

EAR, NOSE AND THROAT

The generalised underproduction of body fluids leads to several problems. Saliva is sparse, causing problems with chewing, tasting, and swallowing foods. The mucous secretions of the nose are excessively thick, forming a crusty mass. Nasal infections are common. A hoarse, raspy voice is common. Abnormal ear wax production may be noticed in some people with ED. The most frequent problem is accumulation of wax in the ear canal. Hearing loss may occur secondary to impacted wax or to nerve degeneration.

EYES

Tears are reduced, causing irritation of the eyes, conjunctivitis, and sensitivity to sunlight. There may be cloudy corneas or cataracts.

RESPIRATORY PROBLEMS

The linings of the nose, larynx, trachea and lungs are moistened by various glands, some of which may be defective in ED. Respiratory problems are therefore common.

For further information about Ectodermal Dysplasia please contact

Mrs Betty Kozanecki
15 A'Beckett Avenue
ASHFIELD NSW 2131
Telephone: (02) 799 9783

SANFILIPPO DISEASE

INTRODUCTION

Sanfilippo disease is a mucopolysaccharide disease and is also known as MPS III. It takes its name from Dr Sanfilippo who was one of the doctors from the United States who described the condition in 1963.

WHAT CAUSES THIS DISEASE?

Mucopolysaccharides are long chains of sugar molecules used in the building of connective tissues in the body.

"saccharide" is a general term for a sugar molecule (think of saccharin)

"Poly" means many

"muco" refers to the thick jelly like consistency of the molecules in solution.

This is a continuous process in the body whereby used materials are replaced and broken down for disposal. Children with Sanfilippo disease are missing an enzyme which is essential for cutting up the used mucopolysaccharide called heparan sulphate. The incompletely broken down mucopolysaccharides remain stored in cells in the body causing progressive damage. Babies and small children may show little sign of the disease, but as more and more cells become damaged, symptoms start to appear.

ARE THERE DIFFERENT FORMS OF THE DISEASE?

To date four different enzyme deficiencies have been found to cause Sanfilippo disease and so the condition is described as type A, B, C, or D. Type A is the most common form found and there are also children with B and C. Type D is very rare indeed and less than ten cases have so far been reported worldwide.

HOW IS THE DISEASE INHERITED?

We all have genes inherited from our parents which control whether we are tall, short, fair, etc. Some genes we inherit are "recessive", that is to say we carry the gene but it does not have any effect on our development. Sanfilippo disease is caused by a recessive gene. If an adult carrying the abnormal gene marries another carrier there will be a one in four chance with every pregnancy that the child will inherit the defective gene from each parent and will suffer from the disease.

HOW DOES THE DISEASE PROGRESS?

The disease will affect children differently and its progress will be much faster in some cases than in others. Change will usually be very gradual and therefore easier to adjust to. The disease tends to have three main stages.

The first, during the child's pre-school years may be a very frustrating one for the parents. They begin to worry as their child starts to lag behind their friend's children in development and they may feel they are being blamed for the child's overactive and difficult behaviour.

The diagnosis is often made very late as some children do not look abnormal and their symptoms are among the most common seen in all children. The doctor has to be perceptive enough to recognise that something serious is wrong and to ask for a urine and blood test to help reach a diagnosis. It is not unusual for families to have had one or more affected children before the diagnosis is established.

The second phase of the disease is characterised by extremely active, restless and often very difficult behaviours. Some children sleep very little at night. Many will be into everything and many like to chew hands, clothes or anything they can get hold of.

Language and understanding will gradually be lost and parents may find it hard not being able to have a conversation with their child. Many will find other ways of communicating. Some children never become toilet trained and those who do will eventually lose this ability.

In the third phase of the disease Sanfilippo children begin to slow down. They become more unsteady on their feet, tending to fall frequently as they walk or run. Eventually they lose the ability to walk. Life may be more peaceful in some ways, but parents will need help with the physically tiring task of caring for an immobile child or teenager.

APPEARANCE

Sanfilippo children grow to fairly normal height and changes in appearance may be less than in other MPS diseases. The hair is thick and coarser than usual and their bodies may be hairier than normal. The eyebrows are often dark and bushy and may meet in the middle.

PHYSICAL PROBLEMS

Of all the MPS diseases, Sanfilippo produces the mildest physical abnormalities. It is important however that simple treatable conditions such as ear infections are not overlooked because the behaviour problems make examination difficult.

Sanfilippo children's fingers occasionally become bent over and the arms may not be able to be extended fully. Later on there may be some limitation of movement in the large joints.

Many Sanfilippo children have frequent colds, blocked noses and chest infections.

Many Sanfilippo children have bouts of severe diarrhoea, the cause of which is not fully understood. It is thought that these may be a defect in the autonomic nervous system. This is the part of the nervous system which controls those bodily functions which are usually beyond voluntary control.

Constipation may become a problem as the child gets older and less active and as the muscles weaken.

At a later stage of the disease a number of Sanfilippo children will start to have frequent minor seizures when they momentarily lose consciousness.

Some may be generalised seizures which can be controlled by drugs.

PUBERTY

Sanfilippo children will go through the normal changes associated with puberty.

LIFE EXPECTANCY

Life expectancy is extremely varied. The average life expectancy is around 14 years but some children do not live as long while others will live into their 20s. Occasionally, very mildly affected individuals have lived into their thirties, or in rare instances, their forties.

IS THERE A CURE?

At present there is no cure for any of the mucopolysaccharide diseases. Various experimental methods have been used to try to replace the missing enzyme. Bone marrow transplant has been tried on Sanfilippo patients, but with disappointing results.

For further information about Sanfilippo Disease please contact

Mrs Denise Law
President
6 Azalea Place
LOFTUS NSW 2232
Telephone: (02) 521 6785

SAFDA

The SAFDA Program, Support After Fetal Diagnosis of Abnormality, is a pilot program currently being developed in conjunction with the SAFDA Steering Committee by the:

NSW Genetic Education Program
Level 3, Block 4
Royal North Shore Hospital
Pacific Highway
ST LEONARDS NSW 2065
Telephone: (02) 438 7324
Facsimile: (02) 906 7529

SAFDA

- acknowledges the shock, grief and sense of isolation experienced by those affected by a prenatal diagnosis of abnormality
- offers confidential and non-judgmental support and information from the impact of diagnosis, through the decision-making process to adjustment in the aftermath.

In its pilot stages, the Program can be accessed through the NSW Genetic Education Program, by self or through professional referral, to obtain:

- Existing written resources on parent and health professional perspectives
- Access to coffee/support meetings, available bi-monthly, which provide an opportunity to explore the issues and safely acknowledge the grief of this tragic dilemma
- Referral to genetic counselling services.

FAMILY STORIES

Before our first born arrived I used to try and imagine what the baby would look like as a toddler if it happened to be a boy. That imaginary boy had curly jet black hair just like his father's, and I almost had him booked into Grammar School before he was even born! We had to wait another two years for that little boy as our first born was a lovely little girl, Lucie. Robbie was born on 17th June, 1981. He achieved all his milestones at the right times - crawling, walking, talking, and as a toddler he had a fine crop of curly black hair.

Life seemed on course for Robbie until he reached the age of 3 years when several things concerned us about his development. We were having difficulty toilet training him and he had persistent diarrhoea. He always had trouble getting to sleep and he was starting to display aggressive behaviour, eg he would start playing with blocks and then throw them, he couldn't ever really interact with other children and would hit them. We took him to our local GP who commented that his liver and spleen seemed enlarged and referred us to a local pediatrician. The pediatrician subsequently ordered several tests, X-rays, blood tests, etc. We have only recently discovered that he suspected a condition known as Mucopolysaccharidosis (MPS) and tested Robbie for it (we didn't know what he was being tested for) and

the tests came back negative. We were told our son didn't have what was suspected and that the reason for his condition was unknown when, in fact, he did have MPS. I remember having an overwhelming feeling at the time that something was being missed. Robbie wasn't diagnosed until 3 years later on the eve of his 7th birthday. I don't particularly like to recall those 3 intervening years and all that happened to Robbie as it is very painful. The sleeping disturbances, behaviour problems, mental deterioration, persistent diarrhoea all increased. We nearly went crazy trying to find out why this was happening.

Mucopolysaccharide storage disorders are a rare group of genetic degenerative disorders. They are caused by a lack of a specific enzyme which, in normal individuals, breaks down complex carbohydrate chains called mucopolysaccharides. These chemical reactions take place within the lysosomes, which are structures found in most cells. In affected children, because the carbohydrate chains cannot be broken down, the under graded mucopolysaccharides are stored in the lysosomes. The abnormally stored material accumulated through the body affecting all the organs and causing complications that eventually lead to premature death. Our son has MPS IIIA which is known by the name Sanfilippo. It particularly affects his brain (central nervous system), liver, and skin.

MPS diseases are inherited from healthy parents who have no idea they carry the genetic defect. Sanfilippo disease is caused by a recessive gene. If an adult carrying the abnormal gene marries another carrier there will be a one in four chance with every pregnancy that the child will inherit the defective gene from each parent and will suffer from the disease. By the time Robbie was diagnosed we had another little boy Peter, 9 months, who was subsequently tested and to our great joy was unaffected.

We endeavoured to find out why Robbie would test negative for a disease he would have had from the moment of conception and discovered that some years ago there were several cases of false negatives. The tests are more sensitive now. Even though the diagnosis was devastating regardless of when we were told, we found it much worse before when we did not understand what was causing all the things which were happening to him. The tragedy for children with an MPS disorder is that life is in reverse. They appear normal at birth and by the time the disease manifests itself sufficiently to cause concern, they may be toddlers or older, making it very difficult to accept that in most cases they will die before reaching adulthood.

Parents of MPS children experience feelings of utter helplessness watching their child lose language and skills which were once simple. Robbie used to be able to peel and eat a banana on his own. Now, if given a banana he tries to eat through the skin. He is losing the ability to chew and will eventually lose mobility. His once curly locks have become straight and coarse. We were put into contact with the MPS Society through a Social Worker to whom we will always feel indebted as the contact has been of enormous benefit. The initial feeling of isolation disappeared after talking with other families about their experiences with their MPS children. We exchange practical information which is particularly helpful when dealing with day to day living with an MPS child. It is difficult to understand why families are sometimes not given the

opportunity to contact support groups when they can make such a great difference to that family's handling of the whole situation.

We now try and plan the best way to manage each situation to make Rob's life easier and our lives as normal as possible. Robbie still gives some very good responses and in the appropriate places. He knows he is loved and secure. He has started to say "Ma" and "Da" again which he lost when he was about 4 years old when his language went. It is wonderful when something comes back which you think is lost forever.

For how long we don't know, but it is precious while it is here. Each day with Robbie is a real challenge which we are determined to meet as even though there are many unwelcome changes happening to him, he is still our beautiful little boy.

Helen Brown

* * * * *

JOSEPH'S STORY

Facing the dilemma of your own child having a rare genetic syndrome can often be overwhelming and give one the feeling of being trapped or looking into a black hole with no way out. However, hearing the experience of other families faced with similar circumstances, helps to remove some of the uncertainties and fears that one is faced with when their child is diagnosed with this syndrome.

Joseph has Hypohidrotic Ectodermal Dysplasia. Hypohidrotic Ectodermal Dysplasia affects the sweat and oil glands, teeth, skin, hair and nails, and is only one of the many types of the Ectodermal Dysplasias. He was born at 36½ weeks after a very uncomfortable pregnancy. He was not an attractive baby, he looked totally different from my daughter at birth, they thought that he was overdue. His skin was very bruised, and very dry. He had no hair on him, not even eyebrows or lashes. After a few days he began to peel. His chin was sunken and due to bad nasal congestion, he had problems feeding. He hated being wrapped lightly in a rug and continually screamed until he was unwrapped. I now put this down to him being too hot.

Around 4 weeks he developed a dreadful nappy rash which we could not get to disappear ... he could not wear any type of nappy. Next we had eczema from head to toe; this was put down to allergies. During summer of 87 he spent most of his time in cool baths. His tongue continually hung from his mouth, some people suggested that he may be retarded due to this. He got hot very quickly and we found it very hard to lower his temperature. By 12 months he showed no signs of getting teeth or hair and was still getting fevers for no visible reason. At 16 months he was diagnosed with having Hypohidrotic Ectodermal Dysplasia. This was a bitter sweet experience. The reality of knowing our son had an incurable disorder that we had never heard of and the relief of knowing he had finally been diagnosed and we finally knew what the problem was. We were not just over anxious parents.

There was virtually no information available to us on the ectodermal dysplasias and we did not know of anyone who had an affected child. We were very eager to gain information on this disorder and how to

manage it. After meeting a dentist in Sydney who deals with these children's dental needs, we found out a lot of helpful information. Also, we were given booklets to read and learned different methods of handling Joseph's disorder.

We had to air-condition our house and our car to keep Joseph cool. If going out on warm days he wears wet T-shirts and hats, and we take a spray bottle to wet him. We no longer go out in the middle of the day and never on extremely hot days. Due to his skin being lightly pigmented and easily irritated he always wears a sunscreen, and has a moisturising lotion rubbed on. We never use soap on him or to wash his clothes as he comes out in rashes. Joseph's hair is dry and unruly due to absent or poorly developed oil glands ... so it only has mild shampoos and lots of cream rinses applied to it. He has only one tooth which is pointed and conical in shape, so he wears dentures. Joseph received his first upper denture at two years of age, and his lower denture will soon be fitted. He has a lot of nasal problems with excessively thick mucus causing foul nasal smells, infections and bad nose bleeds. To try to overcome this problem we have a room humidifier on in his room every night. Also, he has respiratory problems such as asthma and is on daily medication for this. The biggest worry is monitoring his temperature and ensuring that he remains cool. It can be a frightening experience when his fevers become very high and seem uncontrollable; but he is a bright child with a great outlook on life and seems to be coping very well. We will always allow Joseph to make his own decisions on what he can and can't do as he gets older and understands his body cooling problems.

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 Executive Director 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-judgemental approach is recommended in establishing contact.

* * * * *

Mrs I Testolin
1 Ians Grove
Lower Templestowe VIC 3107
12-3-92

The Executive Director
AGSA
Suite 18, 12 Tryon Road
Lindfield NSW 2070

Dear Mary

Thank you for your letter and the 2 back-issues of the Newsletters, I enjoyed reading them very much.

Could you please place my name and address in the "Contact Corner" of your next Newsletter.

My daughter "Leanne" is 18 and has the condition called "KRABBS LEUKODYSTROPHY". I would like to make contact with other families with this condition OR with the condition called "METACROMATIC LEUKODYSTROPHY" as I believe this to be basically the same as the "KRABBS" LEUKO with only a small difference.

Looking forward to reading further issues of your Newsletters.

Regards

* * * * *

16 Strutton Ground • London SW1P 2HP
Tel 071-222 2695 • Fax 071-222 3969

Ms Mary Ford
AGSA
Suite 18, 12 Tryon Street
LINDFIELD NSW 2070
AUSTRALIA

March 24 1992

Dear Mary,

The Cockayne Syndrome Contact Group is a very small group which helps and advises the families of children with Cockayne Syndrome, a very rare metabolic disease.

Mrs Khan, who co-ordinates the Group, has collected much useful information on the syndrome and would be interested to hear from the families of children with the syndrome in other countries in order to develop support and exchange experience to their mutual benefit.

I would be most grateful if you would publicise the existence of the Group in your newsletter and inform any individual contacts about Mrs Khan and the Group.

Yours sincerely,

Diane Barnett
Development Team



The mother of a baby who died shortly after birth from FRASER syndrome would like contact with other parents of FRASER syndrome children.

* * * * *

Request for contact by a mother of an 11 year old boy, James, who has SOTOS syndrome.

* * * * *

The family of a 3 month old baby with a deletion of the short arm of Chromosome 10 would like contact with anyone else with the same diagnosis.

* * * * *

ANGLEMAN (Happy Puppet) SYNDROME

If you are interested in starting a support group for those with this syndrome, or would like contact with a family, please contact

Jenny Chippendale
33 Dean Street
West Pennant Hills NSW 2125
Telephone: (02) 484 6310

* * * * *

Contact required for a family with a diagnosis of Deletion of Chromosome 15

* * * * *

Genetic Counsellor in Tamworth requests contact for a family with a boy aged 12 months who has FANCONI ANAEMIA. Please contact

John Rae, Genetic Counsellor
Community Health Centre
180 Peel Street
TAMWORTH NSW 2340
Telephone: (067) 66 2555
Facsimile: (067) 66 3946

* * * * *

A family have recently had a child diagnosed with Deletion of Long arm Chromosome 10 and would like contact with another family who have had the same experience.

* * * * *

If any of your reader have children affected with METHYLMALONIC, PROPIONIC, ISOVALERIC, GLUTARIC ACIDEMIAS or other acidemias/acidurias, please let them know about our organization —

ElizaBeth Webb Beyer
Organic Acidemia Association
522 Lander Street
Reno, Nevada 89509
Telephone: (702) 322 5542
Facsimile: (702) 323 3869

* * * * *

If anyone has, or knows of someone with SARCOIDOSIS, please contact

Jennifer O'Grady
19 Pegasus Street
RICHLANDS QLD 4077
Telephone: (07) 393 0510, (07) 372 2904

* * * * *

If you would like contact with a family who has a child affected by Sprintzen Syndrome please call

Linda Lawrie
Telephone: 773 3474

* * * * *

SEMINARS/WORKSHOPS

MAKE A NOTE IN YOUR DIARY NOW!

GROUP LEADERSHIP AND COUNSELLING SKILLS SEMINAR

Date: Sunday 23rd August, 1992

Times: 9.30 - 4.30

Venue: Dougherty Centre
7 Victor Street
CHATSWOOD

9.00: Registration

9.30: Outline of day's activities:
Welcome

9.45 Presentation by Jan Grant

WORKSHOP OUTLINE

Group Leadership

Qualities of good group leaders

Stages in groups and what this means for group members and leaders

Task and maintenance in groups

11.00 • • • Morning Tea • • •

Task and maintenance in groups (*continued*)

Using resources and referral sources

How does the leader get her/his needs met and look after self?

Counselling Skills:

1. In the group situation and on the phone or one-to-one

2. Qualities of good listeners:

Non-judgemental

Accepting

Comfortable with feelings

Able to reflect what other is saying

Appropriate use of questioning

Aware of own limits

Sharing appropriately

3. Unloading my feelings — how/where?

PRACTICE

4. Question time

1.00 • • • Lunch • • •

2.00 Small Groups

4.00 Debriefing and general discussion

4.30 Finish

For participants to gain the most benefit from a Seminar such as this it will be necessary to limit numbers. However, if we receive a good response we will endeavour to hold another one as soon as possible

For more information, or to register, please contact:

Mary Ford

Phone: (02) 416 0647

Understanding Maternal Grief

Grief Counselling and Therapy Workshop
with

Margaret Nicol

B.Sc. (hons), Dip.Ed., M.Psych. Clin., M.A.P.S.

Margaret brings her many years of experience, psychological understanding and clinical skills to the workshop. This will be mainly an experiential workshop appropriate for those who have attended her previous workshop or for new attendees. The main focus being on the development of grief counselling skills so that participants may gain:

1. The psychological understanding of pregnancy and the mother infant bond.
2. The impact on women's health of the breaking of the bond through: Miscarriage, Termination, Adoption, Stillbirth, Neonatal Death, Sudden Infant Death, The Birth of a Handicapped Baby or Infertility.
3. KEY FACTORS in healthy grief resolution.
4. Clinical indications of unresolved grief.
5. A Self Psychology approach to grief therapy.
6. Overseas developments in health care innovations.

DATE AND VENUE

A ONE DAY WORKSHOP FOR HEALTH CARE PROFESSIONALS COMMUNITY SUPPORT GROUPS AND BEREAVED PARENTS

DATE: **Friday AUGUST 21st, 1992**

TIME: **9.00 a.m. — 4.30 p.m.**

VENUE: **YWCA, 5-11 Wentworth Avenue, DARLINGHURST (Opp. Hyde Park)**

COST: **\$95.00** (Reduced prices for students and members of voluntary support groups)

CONTACT: **JON GREGORY on 327 86020 or MARGARET NICOL on 362 4790**

For registration make cheques payable to GRIEF WORKSHOP and send with name, address and phone numbers to
JON GREGORY, Suite 7, 1 Knox Street, Double Bay NSW 2028

Receipts will be forwarded. Registration is required 10 days prior to workshop to facilitate organisation.

— • STOP PRESS • —

Have you heard?

The gene for
LOWE'S SYNDROME
has been FOUND!

RESOURCES & Aids



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**Friday September 11th
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Fax: (03) 818 2967

or write
PO Box 493
HAWTHORN 3122

DISABILITY Aids

AND TECHNOLOGY

EXHIBITION

* * * * *

BOOK REVIEW

HOW TO BE PARENTS OF A HANDICAPPED CHILD — AND SURVIVE

Kerry Kenihan
Penguin Books Aust 1991

One in ten Australians is disabled, yet support for their families remains inadequate. The needs of the handicapped child often lock the family into a cycle that strains relationships, resource fullness and dedications.

Sharing the experiences of other parents of disabled children, Kerry Kenihan discusses the problems, self doubts, frustrations and fear of their situation. Her book offers sound practical advice on problems with as professional help, family adjustment and the special difficulties of migrant and single parents, as well as a comprehensive directory of sources of assistance.

This is a book not only for the parents, family and friends of disabled children, but for all those who want to understand them.

* * * * *

RECREATION AND SPORT

'Information For People With a Disability' is a pamphlet produced by NICAN INCORPORATED. Information for all around Australia on a huge range of sporting activities from chess to cross country skiing. Qualified well caring people to help with any disability.

For more information please contact:

The NSW Sports Council for the Disabled
State Sports Centre
Underwood Road
HOMEBUSH 2141
Telephone: 763 0155

*Reprinted from 'In Touch; — The quarterly newsletter of the
Spinal Muscular Atrophy Support Group (QLD)*

* * * * *

INCONTINENCE Aids

Parents occasionally ask about sources of medium and large disposable nappies. United Suppliers, 14 Kendall Street, Granville — Tel: 637 8666 make a large, thick disposable nappy suitable for older children and night time use. They also produce a smaller, thinner version which would be suitable for the children who are between 'Snugglers 3+' and Adult sized incontinence pads. Some families may be eligible for PADP funded nappies. Find out if you qualify by phoning Hornsby or Parramatta Hospitals and ask for PADP.

For people who prefer terry towelling nappies, these are now available in large sizes from the
Corrective Services Department
Unit 1 and 2, 391 Park Road
Regents Park
Tel: 644 4966 or 644 4772.

Parents needing small to large Pilchers can obtain them from

Sunny Textiles
561 Harris Street
Ultimo
Tel 281 2133

They have sizes 0-16 and large ones are \$3.00. They will post them but there is a post and handling charge of \$3.80 per package.

*Courtesy of 'Karonga Courier' 29th April, 1991
(Newsletter of Karonga School, Epping NSW)*

* * * * *

BEREAVEMENT/GRIEF COUNSELLING SERVICES

1. Grief support — Ph: 489 6644
2. Bereavement Care Centre — Ph: 569 9311
3. Compassionate Friends — Ph: 267 6962
4. Grief Line (Calvary Hospital) — Ph: 588 5367
5. Lifeline — Ph: 264 2222
6. Canteen (for teenagers with cancer) — Ph: 399 4604
7. Canya (for young people 21-35 with cancer)
— Ph: 438 7354
8. Sands (Stillbirth and Neonatal Death Support)
— Ph: (02) 450 1565