Please do not remove - File Copy

A G S A

NEWSLETTER

APRIL 1989 ISSUE NO. 1

THE · ASSOCIATION
OF · GENETIC · SUPPORT
OF · AUSTRALASIA

In This Issue

EDITORIAL

The first edition of the AGSA newsletter has finally got to print! Although progress to date has been slower than all who are involved would have liked, I am pleased to be able to report that our constitution has been accepted by the Department of Corporate Affairs and AGSA is now incorporated. Charitable registration is proceeding.

Whilst many of you are aware of the aims of AGSA, I will take this opportunity to emphasise the importance of many voices raised in unison to achieve these goals. AGSA was established to tackle the problems common to families affected by genetic disorders. While many self-help groups exist for the support of families with specific genetic disorders (some 50 in Australia), these disorders are often rare and thus the groups are inevitably small. AGSA will not interfere with the autonomy of the individual support groups but it would obviously be advantageous to join together to increase knowledge and services for families and individuals affected by genetic disorders.

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.

Until AGSA can support an office of its own, any enquiries can be directed to the offices of the NSW Genetics Service in Hut T of the Prince of Wales Children's Hospital at Randwick (telephone (02) 399 2788). Information about all the existing support groups can be obtained from this office and Ms. Mandy O'Reilly, Assistant Co-ordinator of Genetics Education for NSW, can be contacted Wednesday, Thursday and Friday for any enquiries.

At present, the financial resources of AGSA are limited but some of the activities proposed to achieve the aims stated above for member support groups and individuals, include:

- assistance with educational activities of support groups such as newsletter and pamphlet production
- assistance with conference and seminar expenses which may include travel to national and international meetings
- publicity of support groups and individuals affected by rare disorders through the AGSA newsletter
- identification of funding resources for individual groups
- assistance with publicity of particular genetic disorders e.g. the Neurofibromatosis Society has been provided with Media mailing lists for publicity of their forthcoming Awareness Week and Dr. Barlow has offered to speak on their behalf if necessary
- advice on setting-up and incorporation procedures for new support groups and development of educational material

However, individual support groups do not address the problems of genetic disorders as a whole. AGSA proposes, in the short and long-term, to:

- establish a bi-annual bulletin (which is not the AGSA newsletter) to be sent to medical practitioners and other health professionals which will contain updates on genetic disorders (which may include extracts from support group newsletters) and genetic research. This will be a major commitment of funds but is essential to achieve the aim of education of health professionals about genetic disorders
- provide a contact point for individuals affected by very rare disorders through the correspondence column of the newsletter and access to international support group information as well as general advice and support
- publish an information pamphlet about AGSA

to be mailed out to medical practitioners and other health professionals so that AGSA will be a referral source for affected individuals to support groups where they exist and to genetics services

- AGSA may organise it's own conferences and seminars on aspects of genetic disorders relevant to a wide number of support groups and health professionals
- establish affiliation with international umbrella groups for genetic disorders
- join organisations which address health issues to ensure genetic disorders are considered when health planning is formulated.
- raise funds to provide AGSA with a Secretariat and to cover the cost of production and mailing of the bulletin, newsletter and other educational material
- preparation of submissions to Governments to ensure genetics services and relevant community resources are equitable and adequate throughout Australasia.

The Executive is approaching Service organisations and other funding bodies for initial financial support and is also currently preparing a prospectus to put before Corporate Australia for more substantial funding for the on-going activities of AGSA. The allocation of this funding to support groups or individuals will be determined by the Executive Committee elected from the AGSA membership so that all members will have equitable access.

Our membership includes 38 individuals and the following groups:

Friedrich's Ataxia Association

Williams Syndrome

Australasian Tuberous Sclerosis Society

Osteogenesis Imperfecta Society of NSW

The Neurofibromatosis Association of Australia

Spinal Muscular Atrophy Support Group

Dystrophic Epidermolysis Research Association

The Society for Mucopolysaccharide Diseases

Turners Syndrome of NSW

Retinitis Pigmentosa Society

Coeliac Society of NSW

Spina Bifida Group

Cornelia de Lange Syndrome

Little People's Association of Australia

Fragile X Support Group.

The numbers are still small and I hope that many more will join in our efforts by sending in the membership subscription form at the back of this newsletter. The fact that we represent a large number of support groups will be an incentive to sponsorship and help us to achieve our aims.

The present Executive was elected at a meeting held on 15th November, 1988, but due to the requirements of incorporation, the 1st General meeting will be held on May 31st, 1989, at the Paediatric Lecture Theatre (Basement Level), Prince of Wales Children's Hospital, High Street, Randwick 2031.

The meeting will commence at 8.00p.m. with refreshments served at 7.30p.m. All positions will be declared vacant and a new executive elected.

We hope to have a large attendance at the meeting and would appreciate an indication of intention to attend (R.S.V.P. Mandy O'Reilly, Tel. (02) 399 2788).

Your input and suggestions for the further development of AGSA at this meeting will be of great value.

Kris Barlow, *President*

INTERNATIONAL NEWS

We look forward to increasing our contacts with similar umbrella organisations overseas. We are currently aware of the Southern African Inherited Disorders Association, the USA Alliance of Genetic Support Groups, the National Organisation for Rare Disorders and the U.K. group called Contact A Family.

It is hoped that AGSA will work towards improving and increasing access to genetic services throughout Australasia and ensuring that these are on a par with services provided in other countries. Contact with these and other relevant organisations will help stimulate the development of AGSA and provide an opportunity to develop international links. This has particular relevance for rare genetic disorders in a country with Australia's relatively small population.

Letters to the Editor

Dear Editor,

I want to express my delight both personally and as President of the MPS Society at the progress AGSA has made. As a member of the Working Party it has been rewarding to see the enthusiasm all the small Genetic Support Groups have brought to the task of launching AGSA.

As a Society, we have long been aware of the need "to make more noise" about the issues that arise for families with genetic disease. We are a small group (approximately 100 members) and this had always seemed impossible but now I feel that we really have access to a means for lobbying and gaining general information.

I am also convinced that as groups we duplicate each others efforts in many areas. How about sharing information and resources (i.e. workshops) on things like "How to function and administrate more efficiently as a group", "Stress in the family", "Respite Care", "Communication with Professionals", "Loss and Bereavement" and I am sure there are many others.

Anyway, congratulations on a successful launch and best wishes for a bigger and better future.

Yours sincerely,

ROS SMITH, President, MPS Society,
44 Rawson Place, EPPING 2121

Dear Ros,

Thank you for your vote of confidence. I am sure that a united voice will go a long way towards improving services that groups provide to members, as well as those provided by government.

I think your idea of combined workshop/seminars is excellent and we should work towards that as soon as possible.

This is a good opportunity for me to thank everyone who helped get AGSA off the ground. Your help has been invaluable and I am confident that we can work well together to achieve our aims.

Any suggestions and/or criticisms that you or any of the support groups have regarding the newsletter would be most appreciated. I would be happy to hear from anyone with ideas from which AGSA can benefit.

Best wishes,

THE EDITOR

Dear Editor,

I have been invited to speak to a local fundraising group on behalf of my support group. I would like to mention my involvement with AGSA and so I am seeking some guidance from you as to the best way to explain this role.

When discussing our group's involvement with AGSA with our members, I realised that I am unsure of our status within AGSA.

Yours sincerely,

JAN CAMERON-SMITH, O.I. Society of NSW
P.O. Box 401, EPPING 2121

Dear Jan,

Thank you for bringing this issue to my attention. I think this does require some clarification as we must be careful not to misrepresent or cross the lines of autonomy for each group.

You are obviously able to speak as the representative of your group but I think we each need to emphasise our groups positions as members of the AGSA organisation, not its representatives. A member of the executive is the appropriate representative for AGSA.

Yours sincerely,

THE EDITOR

Dear Sir/Madam,

Herewith please find my cheque for $5 for membership of AGSA as advertised in the HD Newsletter of November.

I am a community nurse, also conducting Baby Health Clinics, and have some clients with Genetic Diseases and some at risk. I would like to join so that I can keep up with developments and know where to refer my clients, which is often difficult in the country.

Yours faithfully,

SYLVIA MAISH, The Health Centre,
King Street, THE ROCK 2655

CLASSIFIEDS

We would be happy to provide a section of this newsletter for advertising space for companies providing aids or resources which may be of benefit to members. Costs could be used to offset the production of newsletter expenses. Enquiries should be directed to

The Editor, AGSA,
C/- Hut T, Prince of Wales Children's Hospital,
High Street, Randwick, 2031. Tel. (02) 339 2788.

UNDERSTANDING INHERITANCE AND HOW IT RELATES TO YOUR FAMILY'S HEALTH

Dr. Kristine Barlow, NSW Department of Health.

A chromosome is composed of a chemical called Deoxyribonucleic Acid (DNA) and there are 46 chromosomes in a human cell. A gene is a very small section of the DNA in a chromosome. The number of human genes has been estimated at between 10,000 and 50,000.

Genes contain information for body development, function and structure. Change in gene structure (mutation) results in misinformation and may be expressed as a genetic disorder. The mutations are permanent and will be inherited so that a genetic disorder can then be traced in a family over a number of generations.

While many of the disorders are present at birth, others do not appear until late childhood or in adult life. Approximately 30% of patients in paediatric hospitals are due to a disorder with a large genetic component and approximately 1 in 10 of the adult population will have a chronic disease at least partly with a genetic basis.

In some genetic disorders, early diagnosis — sometimes even before the symptoms appear — can lead to specific treatment. With other disorders, a clinical geneticist or genetic counsellor can often predict how likely that disorder will recur in a family. The 'risk' may be high, moderate or low. Genetic counselling can also provide the opportunity for the fullest possible factual discussion about a disorder and enable couples to make well-informed decisions about their possible options.

A guide has been developed to drawing up a family tree so that it may be possible to detect any disorder which is being inherited in a particular family. This information can be taken to the family physician and may be useful in the prevention of genetic disorders.

How to Draw up your Family Tree

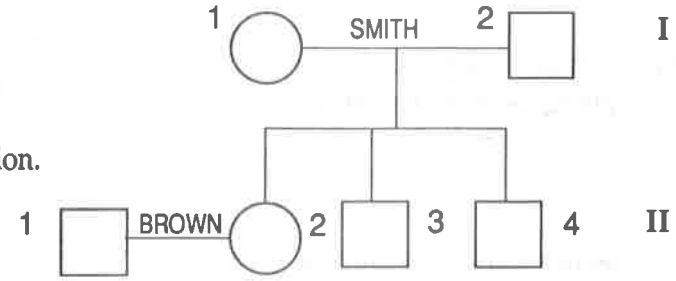
Aim, if possible, for a tree with 3 or 4 generations which you should be able to manage by asking a few questions within your family.

In a family tree, males are represented by

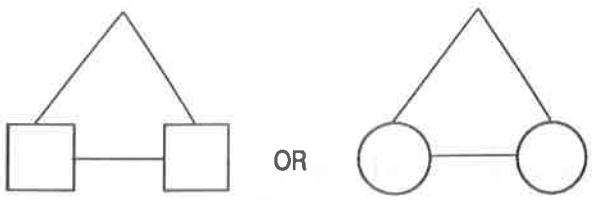
Females are represented by

Each generation is on a separate line with marriage indicated by a line between the male and female. The family name is written on the line.

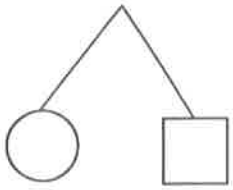
The generations are numbered in Roman numerals and family members given a number from left to write across the generation.



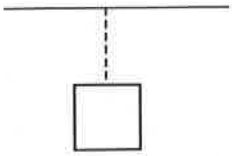
Identical twins are represented by



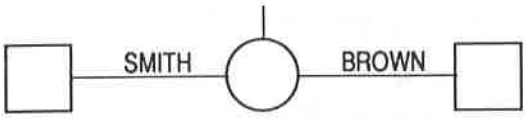
Non-identical twins are represented by



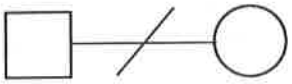
An adoption is shown as



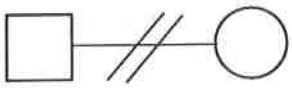
Second marriages are shown as



Separation is

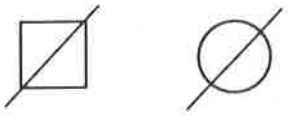


Divorce is shown as

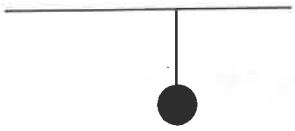


Any deaths in the family should be recorded as they may be important in identifying how an illness has been inherited. It is useful to know when the family member died and what was the cause.

Death is recorded as a cross through the square or circle with the age at death recorded

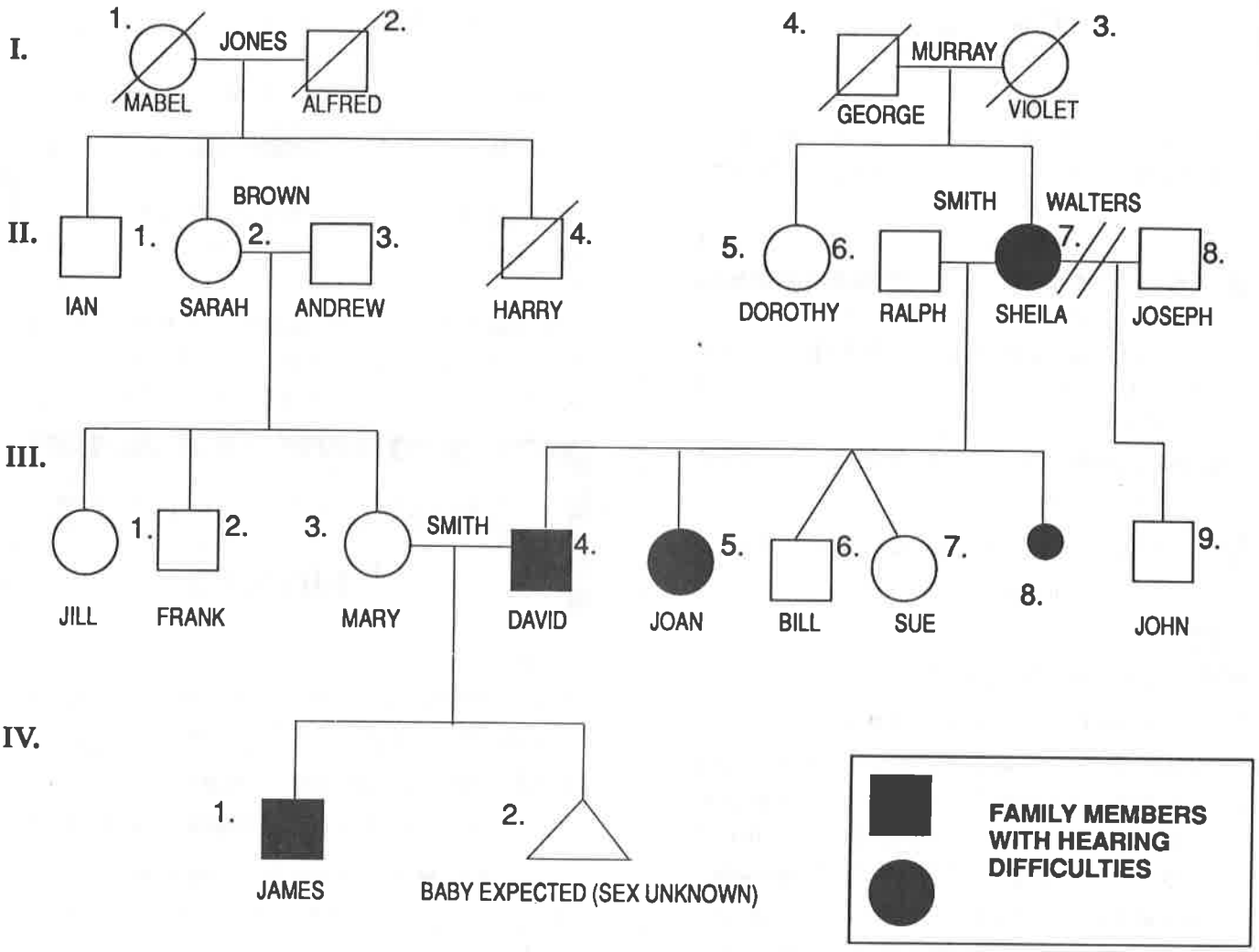


Miscarriages should be noted and are shown as



Any family member who has or has had a medical problem should be identified in the family tree by shading as shown

A family with a history of deafness in its members has been used as an example:



Generation Number	Family Name	Date of Birth	Medical Problems	Date of Death (If applicable)
I-1	MABEL JONES	15/02/12	?	10/2/72
I-2	ALFRED JONES	20/12/10	?	05/09/80
I-3	VIOLET MURRAY	10/01/11	?	13/10/84
I-4	GEORGE MURRAY	16/08/09	?	12/05/71
II-1	IAN JONES	02/02/37		
II-2	SARAH BROWN	04/06/35		
II-3	ANDREW BROWN	02/09/32		
II-4	HARRY JONES	11/12/30	HEART ATTACK	04/03/83
II-5	DOROTHY MURRAY	06/02/30		
II-6	RALPH SMITH	13/11/29		
II-7	SHEILA SMITH	04/01/33	HEARING DIFFICULTIES	
II-8	JOSEPH WALTERS	05/06/32		
III-1	JILL BROWN	02/11/63		
III-2	JACK BROWN	03/11/60		
III-3	MARY SMITH	12/12/65		
III-4	DAVID SMITH	13/05/64	HEARING DIFFICULTIES	
III-5	JOAN SMITH	13/10/66		
III-6	BILL SMITH	03/03/60		
III-7	SUE SMITH	03/03/60		
III-8	MISCARRIAGE	/01/59		
III-9	JOHN WALTERS	15/08/57		
IV-1	JAMES SMITH	02/03/86	HEARING DIFFICULTIES	
IV-2	BABY EXPECTED SEX UNKNOWN			

THE NSW GENETICS SERVICE

The Secretary of the NSW Department of Health has appointed the NSW Genetics Services Advisory Committee to advise on policy development, resource planning and the efficient use of clinical and laboratory genetics services. The Committee is comprised of representatives of all aspects of genetics services (clinical, laboratory and education) and is chaired by Professor Graeme Morgan, Medical Genetics, Prince of Wales Children's Hospital.

Four Clinical Genetics Units have been established at:

Prince of Wales Children's Hospital, Randwick

Royal Alexandra Hospital for Children, Camperdown

Westmead Centre, Westmead

Western Suburbs Hospital, Newcastle

These Units are responsible for genetics services at other metropolitan hospitals (satellite clinics) and in rural areas (outreach clinics) and are at the centre of a network of collaborating clinicians and laboratories.

A Clinical Genetics Unit comprises the staff and facilities required for the provision of clinical and laboratory genetics services and effective interaction between them. Staff include clinical geneticists and Training Fellows, genetic counsellors, laboratory personnel and secretaries.

A clinical geneticist is a medically qualified person, specially trained and HGSA certified for the provision of clinical genetics services. Duties include direct patient contact for diagnosis and counselling, telephone consultations with system specialists, literature review, correspondence and involvement in education, health promotion and research.

Genetic counsellors are graduate health professionals who are involved in clinic co-ordination, genetic counselling with the clinical geneticist, follow-up of families and individuals to ensure counselling has been effective as well as involvement in genetic education and health promotion. Laboratory personnel may be concerned with bichemical, cytogenetic or DNA testing.

The NSW Genetics Service has an administrative base in the NSW Department of Health comprising an Administrative Assistant and Co-ordinators of Genetic Education and Promotion.

The Administrative Assistant is responsible for the co-ordination of the activities of the Genetics Services Advisory Committee and collation and analysis of data relevant to genetics services.

The Co-ordinators of Education and Promotion are responsible for developing programs of public education and health promotion for genetics and for the planning and the implementation of educational programs to ensure that all health professionals may be well informed about medical genetics and kept up to date on important developments in genetics.

GENETIC COUNSELLING SERVICES

Genetic counselling services are available at most major teaching hospitals in Australia.

In NSW, four major genetics units exist:

Prince of Wales Children's Hospital, Randwick.

Royal Alexandra Hospital for Children,

Camperdown.

Westmead Hospital.

Western Suburbs Hospital, Newcastle.

Other clinics are staffed by the members of these genetics units and include:

King George V Hospital

Royal Hospital for Women

Liverpool Hospital

Royal North Shore Hospital

Tamworth Base Hospital

New clinics are constantly being established and further information about the availability of genetics services in NSW can be obtained by contacting Dr. Kristine Barlow, 217 6666.

Other states also have a comprehensive service. Major clinics are held at:

The Murdoch Institute, Royal Children's Hospital, Melbourne

The Adelaide Children's Hospital

The Mater Mother's Hospital, Brisbane

The Princess Margaret Hospital, Perth

The clinics at these hospitals will provide further information about the availability of other genetics service in each State.

GENETIC REGISTERS

The Medical Ethics Committee of the NH&MRC (National Health and Medical Research Council) is examining the establishment and management of genetic registers. A genetic register is a list of individuals who are affected by particular genetic disorders. Following discussion, the Committee will issue ethical guidelines for the research institutions and individuals who establish or maintain genetic registers.

Genetic registers may be established with five objectives, these not being mutually exclusive.

Clinical: to facilitate the follow up and recall of individuals for therapeutic reasons, or when relevant preclinical or prenatal tests become available.

Reference: to store clinical and laboratory data on individuals with genetic disease, so that a diagnosis in a new case might be confirmed by reference to previous cases.

Monitoring: to assess the results of genetic counselling and prenatal diagnosis.

Research: to store and analyse data, or to determine the natural history of particular genetic disorders.

Preventive: to improve the detection and follow up of individuals at risk of transmitting a serious genetic disorder to their offspring so that they may be offered genetic counselling and prenatal diagnosis where possible.

While a register is extremely important in terms of research and family studies, there are a number of ethical issues which require examination and comment. The NH&MRC is calling for submissions from interested groups in this area. Since the advertisement is only being placed in the Commonwealth Gazette, it is unlikely to have come to the attention of most genetic support groups.

If any group is interested in making a submission to the Committee on this subject, please write to:

Ms. Kim Farrant,
Secretary,
Medical Research Ethics Committee,
Department of Community Services and Health,
6th Floor, Albermarle Building,
P.O. Box 9848,
CANBERRA CITY 2601

CONTACT COLUMN

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 Editor may be contacted for the information to be passed on in the case of anonymous requests.

Privacy and anonymity will be ensured if requested.

* * * *

Mrs. J. Loes from the Early Intervention Centre, 39 Geneva Crescent, Wagga Wagga, writes to recommend the American Magazine, "Exceptional Parent" (see review). She also advises that the Adelaide Children's Hospital has been granted government funding for the research of Treacher-Collins Syndrome and requests that anyone who could help out contact her or Sr. Jane Fremantle at Adelaide Children's Hospital, North Adelaide, 5066. Tel. (08) 267 7000.

* * * *

We have a family with Nieman-Pick Disease Type C, Juvenile Onset, requiring contact and information

* * * *

Andrew Murray, 11 McRae Street, Tamworth 2340, (067) 65 4934, has extensive scalp problems and hypoplasia of fingers and toes. He has advised the

Tamworth area Genetic Counsellor, John Rae, that he would like to make contact with fellow sufferers and offer support and information.

* * * *

Can anyone provide information or a contact point for a family with 19q+ chromosomal abnormality?

* * * *

A Medical Geneticist in New Zealand has written on behalf of a family seeking contact with others affected by Pendred Syndrome.

* * * *

Mr. and Mrs. Elliot, Lot 914, Bell's Line Road, Bilpin, 2758, would like contact with any other chronic sufferers of Intestinal Psuedo Obstruction.

* * * *

Noonan's Syndrome? Can anyone help with this request for contact.

* * * *

Is there contact or information for a Muscular Myelopathy affected family?

* * * *

We have printed a request published in the "New Idea" Magazine on 17 September 1988.

"In 1982 a group was established in Victoria called F.R.I.E.N.D.S. (Friedreich's Ataxia and Neurological Disorder Syndrome). This group dissolved three years ago, and Wendy my sister (now deceased) took on the job in 1987 to try to generate new interest. She had just started to get things moving when she died at her wedding in January this year. I have since taken over her files on the group and would like people with the disease to contact me.

Darren Keeble, R.D.D. 70, Boort, Vic. 3537."

The Editor has passed this information on to F.R.I.E.N.D.S. in NSW.

SUPPORT GROUPS NEWS BULLETIN

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

* * * *

President Reagan has declared May 1989 to be Neurofibromatosis Awareness Month in the United States. The Neurofibromatosis Association of Australia aims to use this Public Awareness Month to reach all sufferers of NF, their Doctors and supporters throughout Australia and offer them a support group for sharing ideas and helping individuals. They aim to make the community aware that support and funding are needed to provide literature and information for members and to establish branches in each State of Australia.

In Australia, May 8 to May 14, 1989, is Neurofibromatosis Public Awareness Week and 100,000 information leaflets are being distributed throughout Australia. Copies are available on request from the NF Association

* * * *

Ehlers-Danlos National Foundation USA, Second Annual Conference, August 6-9, 1989. Chesapeake, Virginia, USA.

* * * *

MPS (Mucopolysaccaride Society) are holding the 3rd National Conference at Valla Park Beach Resort, Nambucca Heads from September 15-22, 1989.

* * * *

Obsessive, Compulsive-Neurosis Support Group has now a NSW contact point C/- Mr. Colin Rivett, NSW Association for Mental Health, Information Officer, 62 Victoria Road, Gladesville.

RESOURCES AND AIDS

AGSA does not evaluate or recommend any of the following aids or resources. They are included for information only.

Contact is welcomed from any individuals or organisations who may be able to provide or obtain any information through the AGSA Newsletter. Please send details of your organisation and its aims to the Editor: AGSA, C/- Hut T, Prince of Wales Children's Hospital, High St, Randwick, NSW 2031.

• WESCAP
Western Sydney Citizen Advocacy Program, 7a/70 Macquarie Street, Parramatta, 2150, Tel. (02) 689 2206 or 635 1077. Membership, Groups, $20.00, Individuals $5.00 per annum. Membership is open to all who are interested in the future of Citizen Advocacy in Western Sydney.

• CENTRE FOR EARLY HUMAN DEVELOPMENT
Monash Medical Centre,
246 Clayton road,
Clayton,
Victoria 3168
(03) 550 5470

Multi-disciplinary research unit established to study infertility and the problems of health and development of babies from conception to infancy.

• SPECIAL NEEDS SUPPORT GROUP
55 Moonee Street,
P.O. Box 1977,
Coffs Harbour 2450
Tel. (066) 52 8080

Acts as a network of support for families, provides educational and social gatherings, compiles regular newsletters and administers programs for children with disabilities.

• CASA — Congenital Abnormality Support Association
C/- 7 Loongana Avenue,
Oak Park,
Vic 3046,
Tel. 306-8124.
Aims to provide understanding, encouragement and support to families in adjusting to the diagnosis of any birth defect. To raise community awareness, to liaise with health professionals. To encourage and support data collection and research.

• IDEAS INC. — Information on Disability, Equipment Access Services, Inc.
P.O. Box 479,
TUMUT 2720
Tel. (069) 47 3377
Tell Free 008-029904

Registered charity funded by the Department of Community Services and Health. Has computerised listings for equipment and services for the disabled which are forwarded to the consumer free of charge.

REVIEWS

Please send in any book, article or video reviews you would like to include. This is an excellent opportunity to educate each other and to share information.

"Exceptional Parent" — Published at Boston University School of Education, 605, Commonwealth Avenue, Boston, Mass. 02215, USA.

This magazine has a regular section similar to our Requests for Contact and also includes a Request for Advice column which covers problems such as diet, education, aids, family attitudes and stress which is responded to by other families as well as the Medical profession. Information is also available through advertising for technical aids, resources, courses, conferences and recreational camps. Articles in the March 1988 issue include the following:-

"Improving Social Skills in Schools — the Parents Role"

"Recreational and Leisure Participation — Obtaining Assessments"

"Advice on Camping Holidays"

"Coping with Adolescence in Down Syndrome".

Subscription may be arranged by contacting the Publisher at Boston University School of Education at the above address.

* * * *