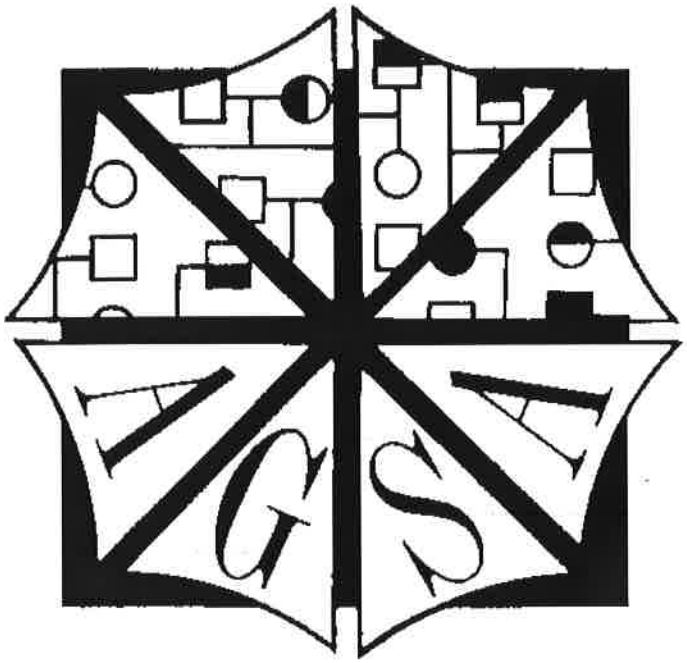

THE ASSOCIATION OF GENETIC SUPPORT OF AUSTRALASIA INC.

FUNDED BY THE NSW HEALTH DEPARTMENT

NEWSLETTER

DECEMBER 1996 ISSUE 27

MISSION STATEMENT

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

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EDITORIAL

We are nearing the end of the calendar year. It has been such a busy year for AGSA, and we are becoming more recognised. It is no longer a concern of AGSA's to find a place in the provision of genetic services, but it is now an essential part of it. I am looking forward to AGSA's future development. On behalf of the committee, I would like to thank the professionals who have contributed their time and expertise at the meetings, for the newsletter and over the phone for Dianne.

The committee has contributed well this year both in some tough decision making and in helping out Dianne AGSA has reaffirmed it's direction, and 1997 is shaping up to be an exciting year.

We will be having a full Awareness Week, not just a day, so we can give many more organisations and families the opportunity to be heard. I asked in my last report for individuals and families who are willing to talk to the media. Only one family was interested. Please think about it a little more!! Every time we have achieved media attention for a specific disorder, new people have been diagnosed and lost souls have been put in contact with each other. If you are interested, please contact the office, and I will discuss it with you.

I would like to take this opportunity to wish you all the best for the coming season. I hope the New Year brings you health and happiness,

From your president,

DEBBIE REDELMAN

PEER SUPPORT/INFORMATION OFFICER'S REPORT

The year for AGSA has been one of unification. Under AGSA's umbrella many people have gathered to share a common goal.

A recent analysis of the people who have contacted AGSA in 1996 (researched by Pam Hutchins, Associate Genetic Counsellor from Melbourne, who worked in my office with me for a week) revealed AGSA is a pivotal point in the genetic community. AGSA has a diverse role in fulfilling the requests of its consumers. It facilitates networking between professionals and consumers in a way which has enhanced communication and understanding of the impact and issues involved in dealing with genetic conditions. AGSA has become a focus for professionals and consumers seeking information, support, counselling and linking others in the same situation. It also has a role in raising genetic awareness in the community through the media and educative seminars.

In November AGSA held an Ehlers-Danlos syndrome seminar . Forty three people attended, ten of whom were younger people with the condition. Many thanks to Professor David Sillence, Maureen Stevens, a Registered Nurse and Dr Jenny Ault who gave excellent talks and demonstrations.

AGSA is planning our 1997 Genetic Awareness Week and we would like to thank the support groups for their response regarding our luncheon forum. I look forward to seeing all the groups together again to celebrate the role of support groups and their organisers.

Many thanks to the people who have returned the AGSA newsletter surveys.

The office will be closed from 25th December to 17th January 1997. I hope you have a happy Christmas and a safe holiday period.

Best wishes for 1997.

DIANNE PETRIE

CONTACT CORNER

AGSA will publish requests for contacts and letters from people searching for families with similar experiences, from those seeking or contributing specific information as well as other resource information.

Anyone who wishes to reply to a request or a letter should write direct to the individual or group concerned where an address is provided. The AGSA office may be contacted for the information to be passed on in the case of anonymous requests. Privacy and anonymity will be ensured if requested.

While AGSA aims to facilitate contacts between families it is unable to assess the suitability of these in individual cases.

It should be remembered that a shared genetic condition does to 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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ANIRIDIA

Sonja Papas, Genetic Co-ordinator, Royal Hobart Hospital, Tasmania, is seeking contact for a family whose two year old girl has this condition. Please contact AGSA or Sonja for details.

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CONGENITAL CYTO MEGALOVIRUS (CMV)

The mother contracted this virus whilst pregnant with her now two year old daughter who was born with brain damage and epilepsy because of the virus. Although it is not a genetic condition AGSA would like to find a contact for this family. Please contact AGSA for details if you know of anyone or have any information that may be helpful.

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GOLTZ SYNDROME

A family is seeking contact for their older daughter who has this condition. Please contact AGSA for details.

LENNOX GASTAUT SYNDROME

A family in Central Queensland would like contact for their 14 year old daughter. Please contact AGSA for details.

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OLIVOPONTO CEREBELLAR ATROPHY

A 56 year old gentleman would like to have contact with another person with this condition. Please contact AGSA for details.

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PSEUDO HYPOTHYROIDISM

A family living in northern NSW would very much like contact with another family.

Please contact AGSA for details.

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RING 22

A mother of a 15 year old boy with this condition would like contact with another family with the view to setting up a support group.

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SPONDYLOMETAEPIPHYSEAL DYSPLASIA (SMED)

A family is seeking contact for their two year son who is one of a twin with SMED. The mother has requested to speak to another family (regardless of the genetic condition) where one twin is affected.

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SUPPORT GROUPS

HEART KIDS FAMILY SUPPORT GROUP

- is a voluntary organisation of parents who have children with heart disease, be it congenital or acquired.

The group offers support to other parents who are currently undergoing a similar crisis, providing the opportunity to share common anxieties and problems. They are simply caring parents who understand what other parents and their families are going through.

A quarterly newsletter is complied to report the activities of the committee and group, raising funds to assist with equipment purchases, research, and various other pieces of information of interest to parents.

The group has information about, hospital stay, hospital Services, What are your social security entitlements and a library of booklets, pamphlets, videos and reference books.

Contact: P O Box 2277, Carlingford Court NSW 2117 or ring Sally on (02) 9796 7918. Lisa (02) 9871 4196, Louise (049) 66 4204, Maree (049) 69-4333, Judy (049) 60 2613.

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CANBERRA PARENTS OF TWINS CLUB

"In Australia, 1 in 73 births is a multiple birth"

The club aims to provide a means of communication for the sharing of information on the care and raising of multiple birth children. Club services include, twin and triplet stroller hire, library, newsletter(TWO-UP), club discounts, clothing pools maternity, triplet adviser etc. Club Activities include, coffee mornings, discussion evenings, family social functions, antenatal information evenings, parents of school-aged multiples (P.O.S.A.M.)

Twin Clubs world-wide are linked through C.O.M.BO, the international organisation

Contact: Robyn Porter Ph (06) 283 2318 (bh)

PROFILE A - Z GENETIC CONDITIONS

It is the intention of AGSA to profile, in each issue, a particular Support Group/Disorder, thus increasing awareness within our membership of the range of genetic conditions. Also it hopes that where overlaps occur in conditions, support Groups may liaise with each other and thus gain a broader understanding of facilities, aids, etc. that may be of value to your individual membership.

Please ensure that all support group information is recent and reliable. It is of paramount importance that you let us know your group is "Alive and Well' and happy to take referrals.

For your information we profile.....

MARFAN SYNDROME

SYNONYMS : MFS

Marfan syndrome is an inherited disorder that effects the connective tissues of the heart and blood vessels (*cardiovascular system).* The musculoskeletal system (*ligaments and muscles)* is also affected. Major signs also include excessive height, long hands and feet, and involvement of the lungs and the eyes.

It was first described by a physician named Marfan in 1896. The diagnosis of Marfan syndrome has been suggested in at least two historical figures, Abraham Lincoln and Paganini.

Symptoms

Symptoms may be severe or mild, and may be present at birth or show up in adult life.

People with Marfan syndrome are usually tall and thin. However the disorder shows a striking variability between affected people, even within the one family. The face, limbs and digits (fingers and toes) are abnormally long. Other features may include excessive joint mobility, flat feet, muscles weakness (*hypotonia)*, a protruding or indented breast bone *(sternum)* and curvature of the spine *(scoliosis).* The teeth may be crowded because of an abnormally high palate. Stretch marks *(striae)* may appear on the skin.

People with Marfan syndrome may have significant cardiovascular problems. The most common of these is mitral valve prolapse, which often occur without symptoms. Mitral valve prolapse is characterised by the incomplete closure of the heart valve and the backward flow of blood in the heart. Enlargement and degeneration of the aorta (the major blood vessel of the body) *aortic aneurysm* (a bulge of the wall of the aorta), and *aortic regurgitation* (backward flow of blood) are also common. Untreated, these cardiac complications account for most deaths from Marfan syndrome.

About 50 percent of people with Marfan syndrome experience an abnormal displacement of the lens within the eyes *(ectopia lentis).* Another major symptom is nearsightedness *(myopia).* Other findings that relate to the eye may include an increased axial globe length, flatness of the cornea and occasionally retinal detachment. These conditions are diagnosed by an ophthalmologist (a physician who specialises in eye disease). Emphysema, which causes destructive changes and the loss of elasticity of the lungs, develops in some patients with Marfan syndrome. A collapsed lung *(pneumothorax)* occurs in 5 percent of affected people, either spontaneously or traumatically, and requires immediate attention.

Causes

Marfan syndrome is a genetic disorder which follows a pattern of inheritance in families called "autosomal dominant inheritance".

Scientists have found that a gene located on the long arm of chromosome 15 (at site 15q15-21.3) contains the information to instruct the body to produce the fibrillin protein, an important protein in elastic tissue in organs such as the blood vessel walls, the eyes, tendons, ligaments and lungs. Everyone normally produces this protein but people affected with Marfan syndrome have a change *(mutation)* in this fibrillin gene. Each

family seems to have its own unique mutation or change in the fibrillin gene.

Everyone who has a mutation in the fibrillin gene will have Marfan syndrome *(penetrance is complete)* but expression of symptoms *(clinical manifestations)*may be variable. Mutations in the fibrillin gene may cause changes in the production of the fibrillin protein, in the way it is deposited or the way in which it is incorporated into tissues.

Research into fibrillin gene mutation is currently in progress at the New Children's hospital, Westmead, Sydney.

About 25% of cases of Marfan syndrome are due to a new mutation of the fibrillin gene. In many of these cases the chromosome carrying the faulty gene will have come from the father. Thus these people will have had no family history of the disorder but will then be able to pass on the mutation to their children. These cases are called *sporadic.*

Affected Population

Marfan syndrome affects males and females in equal numbers. It is not uncommon for this disorder to be misdiagnosed or missed.

Therapies: Standard

All affected people with Marfan syndrome should avoid sports, heavy lifting and any exercise that increases the strain on the aorta produced by vigorous beating of the heart. Drugs which reduce the strength and frequency of the contractions of the heart and which may reduce the strain on the aorta, have proven useful in treating the cardiovascular symptoms. The dosage of these drugs needs to be adjusted to the individual patients needs, and therapy should be closely monitored. However, surgical replacement of the aorta may eventually become necessary.

In the skeletal system, scoliosis and deformity of the chest may represent a serious problem for people with Marfan syndrome. Deformities of the sternum in people with Marfan syndrome (both protruding and inverted breast deformities) may be corrected surgically.

The eyes require careful attention from early childhood. Failure to detect any of the several abnormalities that can affect the eyes may result in a dimness of vision and other visual impairment, increased risk of retinal detachment does demand special attention. The eyes should receive special protection from injury during work or sports. Sports that may involve trauma to the head, such as football, boxing, and diving, should be avoided.

Every person with Marfan syndrome should have regular tests to check the size and function of the heart and aorta. Impaired functioning of the heart valves may responded to various cardiac medications. However, surgical replacement with an artificial valve may become necessary.

As a result of regular surveillance, the use of appropriate medication and improved surgical techniques, the lifespan of individuals with Marfan syndrome has dramatically improved.

Other treatment is symptomatic and supportive.

Genetic counselling is recommended for affected people and their families.

For more information on Marfan syndrome, please contact:

The Marfan Syndrome Association Inc.

P O Box 973

Parramatta NSW 2124

The following overseas groups may be able to provide additional information and support:

Marfan Syndrome Association Inc.

P O Box 973

Parramatta NSW 2124

National Marfan Foundation

382 Main Street,

Port Washington, New York, NY 11050 USA

(516) 883-8712

Marfan Association UK

6 Queens Road,

Famborough GU14 6DH UK

Tel: 01252 547441 Fax: 01252 523585

This information has been kindly supplied by the NSW Genetic Education Program and Dr Lesley Ades, Clinical Geneticist, The Department of Clinical Genetics, The New Children's Hospital.

FAMILY STORY

LYNDA'S STORY

Wow, what a size! These were the words of the doctor, assisting at Anthony's birth. He weighed nearly 5 kilos and was 58 centimetre in length. However, it was not his size that struck us a we proudly admired our second born son, but the length of his fingers! They seemed to go forever - convincing us (as if any proud parents needs convincing of their child's brilliance!), that we had produced a future concert pianist, or perhaps a brain surgeon! All was well with the immediate post birth check of Anthony, by a paediatrician, however, the six week check revealed "clicking" hips and I was instructed to keep two nappies on him at a time. An x-ray at three months showed that all was well and that the double nappies were no longer required.

Anthony kept wonderful health as he grew up, so visits to doctors were few. He was chubby and round faced as a toddler and had an enormous appetite. From school age, he became progressively more long limbed and thin. On one occasion our GP at the time mentioned that he could have Marfan syndrome (from his clinical appearance) and he explained it to me in the most simplistic terms as causing the sufferers to be tall and thin, and long limbed, and very few other details. He did not suggest any testing and really mentioned it only as an aside as we discussed the prevailing reason for our visit. I thought little more about it, and given the way it was presented to me, was not worried.

When Anthony was 10 years old, I sought a referral to a paediatrician as a deformity in Anthony's chest was causing concern to his father and I. The paediatrician gave him the all clear and said that should the chest deformity worsen, an operation was possible at a later stage. No mention of Marfan syndrome. The mother in me sensed that there may be a problem and I sought a second referral to a paediatrician. This doctor ordered checks by a cardiologist, orthopaedic surgeon and eye specialist. All of these tests were unremarkable and whilst I was very encouraged by the results, the paediatrician was in no doubt that Anthony had Marfan syndrome. I did a little reading on the condition and convinced myself that as Anthony appeared to have a few of the listed characteristics (apart from the tall thin stature and chest deformity) he perhaps did not have it. I did, however, think of it constantly, and when Anthony was nearly 13 years old the clawing of his toes (present in mild form from birth) had worsened and I felt another trip to the paediatrician was warranted. He had not wavered from his original diagnosis and again ordered the checks by the three aforementioned specialists. This time, the news was not so good, and the cardiological investigation revealed mild dilatation of the aortic root and slight mitral valve prolapse. This, together with the chest deformity, hammer toes, crowded teeth and tall thin stature, left the paediatrician absolutely convinced that Anthony had Marfan syndrome. The cardiologist prescribed beta blockade and Anthony immediately commenced taking atenolol, 25 mg daily.

With this confirmation, Anthony's father and I were initially inconsolable, asking ourselves, futilely of course, why it could not have been us, as his life was just beginning. Given that Anthony is very academically gifted, having been the Dux in his first and second years of high school, that life and the future looked particularly bright. We kept all our angst from Anthony and presented all the information at our disposal to him in a very matter of fact fashion. With the information and reassurances from the medical

professionals we see Marfan syndrome as no great impediment to the future for Anthony, and nor does he. He has accepted without question or complaint the restrictions and exclusions to his physical activities and participates enthusiastically and competently in those sports that are available to him - currently tennis in summer and soccer in winter. He is now in his third year of high school, continuing to excel academically, is handsome (well, if you overlook the braces on his teeth!), tall, and a thoroughly delightful, happy boy, who is involved to the fullest in all aspects of school life. He is in the school guitar ensemble, edits a school newspaper and is in a top debating team, to name but a few. I mention this only to show that Marfan syndrome need not be a handicap to success.

Anthony has elected not to reveal that he has Marfan syndrome to any of his peers (although, necessarily, his full medical details are held by the school) and we respect his right to this privacy. He will know when the time is appropriate to share the information and it is his decision. Neither my husband nor I have Marfan syndrome therefore in Anthony's case it is a new mutation.

We have joined the Marfan syndrome association and my husband I are committee members. We feel very much that there needs to be a greater awareness of the syndrome in both the medical and general community and are committed to working towards this goal. If this awareness were present when Anthony was born, he would have been diagnosed immediately, as many of the clinical signs were there. We are most anxious that awareness of the syndrome be promoted to enable a much earlier diagnosis in sufferers and earlier intervention in treatment and monitoring of the symptoms.

RESOURCES

BOOK:'HUNTINGTON'S THE NAKED TRUTH'

COMPILED WITH THE MUCH APPRECIATED ASSISTANCE OF THE MEMBERS OF "THE GENE FAMILY HUNTINGTON'S SUPPORT GROUP (CENTRAL COAST & NEWCASTLE)'

Based on their own daily experiences of living with Huntington's. This is an educational booklet on Huntington's for all Affected Families, Professionals (Doctors & Nurses) and Services Providers. Recommended Retail Prices $9.95 + Postage & Handling $2.00 (if applicable) Contact: Pete Smith Publications,9/5 Broula Close, Kincumber, NSW 2251.

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AUSTRALASIAN TUBEROUS SCLEROSIS SOCIETY REPORT

During September 1996 an International Research Symposium on Tuberous Sclerosis was held in Bath, England. Tuberous Sclerosis (TS) is a multi-system genetic disease causing tuberlike growths in the brain and frequently in other organs i.e. kidneys, lungs, liver, eyes. Typical symptoms of TS can be one, some or all of the following - epilepsy, intellectual impairment, behaviour problems and skin conditions. TS is carried by an autosomal dominant gene. It is estimated that TS occurs as a spontaneous mutation in 80% of cases. Where one parent has TS there is a 1:2 chance that each child will also have the disease. TS occurs in approximately 1 in 7,000 people.

The International Research Symposium was attended by Sue Pinkerton (President of Australian Tuberous Sclerosis Society) and Lynn McKinnon (Secretary/Treasurer of Australian Tuberous Sclerosis Society) thanks to the generosity of members and friends of ATSS. Donations covered 65% airfares and expenses, the remainder was from the Society's funds. This was the first International Meeting attended by Australian Delegates, although close ties had been established with Associations in Great Britain and America over a 16 year period.

ATSS has a mailing list of 480, 310 are TS affected families. It was generous donations from these members and friends, many small amounts combined with some donations from

community clubs which contributed to the travelling costs of the two delegates The benefits of attending this international meeting included:

* establishing personal links with TS associations in over 15 countries

* sharing information and ideas with delegates in National Associations

* hearing first hand reports of the most recent worldwide genetic research and medical studies

* participating in discussions with the presenters of research papers

Besides these tangible benefits, both delegates gained a renewed enthusiasm and inspiration for TS Society activities.

Sue Pinkerton

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News from the Alliance Newsletter Alert

New on the Net:

The Gene Letter is an Internet-Based newsletter on the scientific and societal issues in genetics. The purpose of this electronic newsletter is to inform consumers and professionals about advances in genetics and to encourage discourse about emerging medical, ethical, legal and policy dilemmas. It is funded by a grant from the US Department of Energy/ELSI Program. The editors are Philip R. Reilly, MD, JD, Dorothy C.Wertz, PhD, and Robin JR Blatt, RN, MPH. The Gene Letter also operates an uncensored chatroom. Check it out: WWW.geneletter.org.

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Klinefelter Study: Brenda Eskenazi, a professor at University of California, Berkeley School of Public Health, and Any Wyrobek, a researcher at Lawrence Livermore National Laboratory, are studying whether the rate of aneuploidy (extra chromosomes) in the sperm of fathers is different, depending on whether the father did or did not contribute the extra X chromosome. They seek families with a son who is less than six years old with a karyotype of 47-XXY. The father, mother and son will be asked to participate in the study. Contact the Klinefelter Study Officer at 510-642-9545 (collect calls accepted).

CONFERENCES

FROM THEORY INTO REALITY -

Genetics and Genetic Support Groups in the 21st Century

* Saturday, April 5 from 8 am - 6 pm at Bradeis University, Waltham, MA. An overview of the new genetics, its technologies and their relevance to support groups, this conference will offer a combination of lectures, panel discussion, and hands-on workshops in molecular genetics, cytogenetics, and computer laboratories. It is directed towards genetic support group leaders, board members, directors, co-ordinators, and anyone else thinking about running a genetic support group. Presented by the Brandeis Genetic Counselling Program and the New England Regional Genetics Group (NERGG) Consumer Concerns Committee. Registration: $25. For more information, contact Barbara Lerner, MS, Brandeis Genetic Counselling Program, Brandeis University, Waltham, MA 02254; TEL: 617-736-3179.

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First Annual Stickler Syndrome Conference * June 6-8; Best Western, Canterbury Inn, Iowa City, Iowa. Contact Stickler Involved People, 53 Angelina, Augusta, KS 67010; 316-775-2993.

Merry Christmas

Association of Genetic Support of Australasia (AGSA) Inc.

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Individual	$20.00
Group/Organisation	$40.00

Subscription Year lst July - 30th June

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

AGSA aims to:

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

* facilitate access to individual support groups for those families with a particular genetic disorder.

* provide a forum for the exchange of information between support groups regarding available community services.

* educate the medical and allied health professionals and the community about genetic disorders.

* consult with government bodies, both Federal and State, for appropriate funding for genetic services.