THE · ASSOCIATION
OF·GENETIC·SUPPORT
OF · AUSTRALASIA

FUNDED BY THE NSW HEALTH DEPARTMENT

MARCH 1995 ISSUE 17

MISSION STATEMENT

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

EDITORIAL

Charity Awareness Week (NSW) 1-7 May, 1995.

As previously mentioned, AGSA will be participating in Charity Awareness Week 1995.

The 1995 Awareness campaign will identify and promote the ongoing work of member charities by production of a Directory, which will be distributed throughout member charity networks, and all NSW libraries and other target organisations. Other promotions planned include posters, community service announcements, and newspaper articles featuring stories about participating charities. Banners will be prominently displayed on Sydney metropolitan freeway sites.

Please note that a church service will be held at St. Andrews Cathedral, George Street, Sydney (next to Town Hall Station) on Sunday, 30th April, 1995 at 10.30am to mark the commencement of Charity Awareness Week. An invitation to attend the service is open to all.

Further, a Symposium dealing with issues such as the respective roles of Government and Non Government agencies, the duplication of services, NSW Government direction on delivering disability and welfare services is planned for Friday, 6th May, 1995 at Parliament House Theatrette, Macquarie Street, Sydney. An invitation from the Patron and flyer outlining the Symposium Program will be sent to all Charities participating in CAW and others, urging board members and Chief Executives to attend.

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

Information obtained from the Symposium will be disseminated to all AGSA membership.

To round off Charity Awareness Week, AGSA has planned a Trivia Night, for Saturday, 6th May, 1995. Information regarding this evening has already been forwarded to you about this. Please endeavour to make a table of 8 and join us for a fun filled night. Those with lots of brains can join me!

Lysosomal Storage Disorder Group

We have received a number of enquiries regarding the proposed Lysosomal Storage Disorder Group, following production of Professor Hopwood's article in the previous newsletter. Further information related to the meeting planned for 29th October will be circulated in forthcoming Newsletters.

We appreciate that many disorder groups involved will need time to discuss this issue with their respective committees and membership, and individuals, where no group exists, will require time to assess the benefits of such an "umbrella" group

We look forward to a very interactive group on 29th October, so please nate representatives to attend this meeting.

I would further advocate that medical and allied health professionals advise their patients of the proposed Lysosomal Storage Disorder Group. If I can assist further with this particular issue, please feel free to ring me on (047) 515 872.

Ros Smith

INFORMATION/ SUPPORT OFFICER'S REPORT

Thank you to the groups who sent in brochures for display during Charity Awareness Week.

On 19th March, AGSA held a Sotos Syndrome meeting. Dr Anne Turner, Clinical Geneticist and Dr David Starte, Developmental Paediatrician, both gave excellent talks. Six families attended and a number of apologies were received from country areas. The meeting was very successful with families exchanging information and sharing the relief of breaking their isolation. It was decided to set up a support group, produce a newsletter, to locate other families and to meet again at the end of the year. These conditions may be rare, so to these families, just meeting another family is a very rewarding and fulfilling experience and, one of the highlights of my position.

The Living Grief Seminar has been finalised and will be held on 29th July 1995. Numbers will be limited to twenty five and a charge of $40.00 for members and $60 for non-members has been decided upon. This includes registration, morning and afternoon tea plus lunch. (See the attached flyer for details).

I am presently assessing AGSA's membership and if your next newsletter has a big red dot on it- you guessed it! We would like to know if you wish to remain members as your membership payment has lapsed.

This year I hope to attend a number of support group meetings starting with the educational seminar on Charcot-Marie-Tooth disorder on 9th April. I look

forward to meeting you and learning about your group.
It has been a year since I started working for AGSA and during that time 99 people have visited the AGSA office and 941 calls have been received. The position has become very varied and challenging and I have enjoyed my year and look forward with interest to the coming months.

So ring me and see what AGSA can do for you.

Dianne Petrie

Coming events ;-
Expressions of Interest

A. Don't forget the trivia night!! 6th May

Greenlees Park Bowling Club, Concord
Cnr Wellbank Street and Ian Parade
Arrive at 7pm for a 7.30pm start

B. Lysosomal Storage Disorders Meeting
29 October 1995
Expressions of interest are invited from those wishing to attend this meeting. Please write to AGSA or telephone Dianne Petrie at the AGSA office. If you are uncertain about this group of disorders please refer to the article by Professor John Hopwood in the February AGSA Newsletter

RESOURCES

Fragile X Syndrome
Updated information, pamphlets and booklets, on Fragile X syndrome can be obtained from
Fragile X Program
C/- Hospital School
High Street
Randwick 2031
Tele (02) 399 2292
Fax (02) 314 5075

The Fragile X Support Group of WA, in conjunction with Hereditary Disease Program, of the Health Dept of WA, has produced an Information Kit for parents of newly diagnosed Fragile X children. For information contact the Fragile X support groups in each state.

Cystic Fibrosis

The Cystic Fibrosis Association of Victoria has produced a Cystic Fibrosis Resource Kit which can be purchased from the Association's Office at 12 Clarement Street
South Yarra 3141
Tele (03) 8261811
Freecall for STD 1800 633 685
Fax (03) 8263437

The Kit outlines the cause, nature, diagnosis, and treatment of CF as well as the psychosocial issues that may accompany the disorder and provides a discussion of recent technological advances and some related ethical issues.
The kit is recommended for use by professionals in the medical and allied health services as well as for those who live with people who have CF.

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.

For your information we profile.....

Sotos Syndrome

kindly edited by Dr Anne Turner, Clinical Geneticist, Prince of Wales Children's Hospital

General

This rare syndrome whcih has also been called "Cerebral Gigantism" was first described by Juan Sotos in 1965. Sotos Syndrome is a condition resulting in developmental delay and large size apparent in early childhood.

The condition is often incorrectly diagnosed. At present diagnosis is made by observing a range of physical and developmental characteristics, supported by X-ray assessment of bone age.

Features

Although there is no diagnostic test for Sotos Syndrome there are a number of indicators of Sotos in affected children of 1 to 5 years of age.

Babies diagnosed as having Sotos Syndrome are generally significantly larger and heavier than average due to excessive pre-natal and post-natal growth.

Craniofacial characteristics in the newborn and young children include a prominent high and broad forehead, a long head (front to back), a long face, downslanting eyes, sparseness of hair at the temples (> 97% in early children), upturned tip of nose, a long pointed chin, a prominent upper jaw, and a highly arched palate. Other features include large hands and large long feet, teeth which sometimes show excessive wear and discolouration (and which erupt earlier than expected), and nails which are often brittle and thin.

Babies diagnosed as having Sotos Syndrome are very poor feeders, with 40% needing tube feeding. Feeding does improve with age and often young children show a greater appetite than other young children.

Babies also display hypotonia or low tone but this also improves with age. Prolonged drooling and mouth breathing may be present due to poor tone of facial muscles.

As children, Sotos sufferers show a delay in speech development, clumsiness and poor coordination. These difficulties are related to gross motor problems more often than fine motor control although both may be apparent. Coordination improves as muscle tone improves.

Receptive language ability, that is, ability to understand others, develops more quickly than expressive language, or formation of words. As a consequence, children with Sotos suffer considerable frustration in communicating with family and friends. Children will sometimes scream in an effort to express him or herself. With age children tend to develop normal speech and communication patterns, aiding in socialisation and learning. Children with Sotos may mature intellectually, socially, and emotionally on widely different timetables.

Many children will experience some seizures, but only about half of the seizures

experienced by toddlers are associated with fevers, that is , are febrile fits.

Children often have flat feet and occasionally scoliosis. Indications are that there may also be an increased tendency to fractures.

Growth indicators are important in the diagnosis of Sotos Syndrome.

Overgrowth is present prenatally.

At birth Sotos Syndrome babies characteristically have a large head with around a half of newborn babies having a head size above the 97th percentile at birth. After birth the head size of affected babies continues to grow rapidly such that by 12 months of age virtually all affected babies have a head size above the 97th percentile.

Growth is rapid in the early years, particularly from 1-5 years with children usually being over the 97th percentile throughout these years. In adolescence and adulthood, height tends to fall towards average for the population, however, final height in boys, particularly, tends to remain taller than average.

Even though affected babies have a high birthweight, children and adults are not usually overweight. Height indicators are much more significant than weight.

Puberty in girls is reached early, the average age at the first period is 11 years and 3 months. Boys may often have a late puberty.

Development

Development in Sotos Syndrome babies and children is slow. Many individuals come to be noticed because of learning problems.

Early reports on Sotos Syndrome suggested mild to severe mental handicap. **These reports were overly pessimistic and overly simplistic.** In fact a very wide range of intellectual potential has been found, and typically, with an uneven pattern of abilities among children with the disorder.

Problems that have been noted include short term memory, difficulties in handling abstract ideas, practical reasoning, and numeracy even where intellectual capacity is normal. Almost all have greater difficulty with numeracy than literacy.

Because very young children with Sotos Syndrome typically have delayed aquisition of specific motor and verbal skills, and low tone, early assessments often suggest low intellectual capacity. However, with age comes improved learning and maturity such that early assessments correlate poorly with later attainments. Behaviour problems and immaturity may also interfere with school achievement, even where intelligence is normal.

X-Ray Features

Growth pattern and bone age are very good markers in the diagnosis of Sotos Syndrome.

Aproximately 84% of young children with Sotos Syndrome have an advanced bone age (in excess of the 90th percentile for bone development at that age). In fact probably all Sotos Syndrome affected young children have advanced bone age, but some may not have been assessed as well as might be possible.

Metacarpophalangeal pattern profiles are used in assessments as these show a very characteristic pattern in Sotos.

X-ray diagnosis is not as effective as diagnosis by experienced clinicians and

mistakes in diagnosis can occur, both false positives and false negatives.

Genetics

Generally cases of Sotos Syndrome are sporadic, that is, occur for the first time in a family.

The mode of transmission of this syndrome is not known but it is thought likely that it is a genetic condition. Sotos Syndrome affects males and females equally.

Advanced bone age is not present in adults.

As the condition was only described in 1965 there is understandably a paucity of information on adult features.

Further information on Sotos Syndrome can be obtained from:

Sotos Syndrome Support Group
C/- Debbie Drayton
11 Lewis Street
Regents Park 2143
Tel (02) 649 3385

Sotos Syndrome Support Association
737 Shandra Street
Ballwin, MO, 63021 USA
Tel (314) 966 4194

Sotos Syndrome Support Group of Great Britain
C/- Mrs Bridget Veitch
Kilndown House
Kilndown, Cranbrook
Kent, England TN172SG.

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FAMILY STORY

CHARLOTTE'S STORY

Our story begins in April 1989. From Charlotte's birth we were aware something wasn't quite right. She was a large baby, very floppy and didn't want to feed.

Once home she didn't develop like our other three children. After visiting our paediatrician we were sent to the Genetics Clinic at Camperdown. Sotos was suspected but it was two years before it was confirmed. Unfortunately, we now knew what was wrong, but, NO INFORMATION. Our paediatrician gave us a copy of the information he had, not a lot, but something at least.

Charlotte's milestones were all delayed, sitting at nine months, crawling at twelve months and walking at twenty-one months. Charlotte is nearly six now and over the past six years we have coped well despite; difficult feeding, unsettled baby, teeth wear and decay, unco-ordinated movements, clumsiness, proneness to sickness, screaming when insecure, late toilet training, very loud cry and laughter, delayed development in both physical and speech areas, and Metatarsus Varus Deformity (a treatable foot problem). I've no doubt I have missed a number of things out.

At two years and two months we began having Charlotte assessed for the Early Education Program and at two years eight months she began. Over the next three years Charlotte attended Early Intervention Programs as well as home programs and has now started in a regular kindergarten. We are happy with her progress but are working steadily to keep her development on a par with her kinder class.

Our three other children had to grow up quickly as Charlotte took so much time. We have tried to make sure they haven't suffered. Although Charlotte's difficulties are not as physical anymore it is just as demanding.

I had concerns about the affect of Charlotte's birth on us as a family. Our eldest daughter said it all very clearly in an English assignment in Year 8. She stated the happiest day of her life was when Charlotte was born.

Charlotte is a very special little person, she brings a great deal of joy and laughter into our lives. We have had many problems over the past six years and at times I have wanted to give up, but each time Charlotte smiles at you with big brown eyes and gives her funny little laugh, you know that you'll keep working to make her life very special and to ensure that all of our children get the very best quality of life we are able to give them.

MY SON NATHAN

My son Nathan is a text book Sotos child with all the typical symptoms including large hands and feet, large head with distinctive facial features, low muscle tone, accelerated growth, high palate, short sightedness, poor speech capabilities and a developmental delay.

Nathan's life started with prolonged jaundice, leaving hospital after two weeks and returning one week later because he was failing to thrive due to poor feeding. Nathan began having physiotherapy at three months of age because of low muscle tone. There was no way he could hold up his oversized head. He also had a lot of problems with colds, chest infections and middle ear infections and always seemed to be visiting the doctor. At this stage Nathan hadn't been diagnosed as having any particular syndrome and I was told by the paediatrician at the time I should not worry as he was performing within the normal range. I had my doubts but trusted the paediatrician.

When Nathan was one year old he was rushed to hospital with severe croup. The attending paediatrician was immediately aware something was wrong with Nathan and had the Developmental Disability Team visit us that afternoon. From that point on Nathan started an early intervention program consisting of speech therapy, occupational therapy and physiotherapy. We also started attending a DD group where finally I wasn't alone with my special child. What a great feeling it was to talk to other parents whose child also needed extra help.

What a lot of work these first few years were, where sometimes it took weeks or months to make the smallest progress. But when he made that small achievement that most parents take for granted, everyone heard about it. How proud I was of Nathan and myself at these times.

Nathan started at a wonderful preschool when he was four years old and after the initial problem of him not wanting to leave his mum, he fitted in very well and loved the social contact with other children his age. Nathan's strong point was gross motor which meant he had very little problems in keeping up with his peers in the playground. Nathan loved kicking balls, riding bikes etc, and although his language was poor he could nearly always get his point across. I received a lot of support and encouragement from the preschool and we all agreed it would be best for Nathan to concentrate on the social aspects to prepare Nathan for school. By the second year at preschool Nathan was participating in everything and had quite a few friends who seemed to look up to Nathan.

Nathan's health was still rather poor due to constant colds and ear infections and had two lots of grommets in his ears and his adenoids out. Rushing to hospital with severe croup became fairly common as well as visits to the podiatrist for insoles for his shoes due to flat, rolling feet. Hearing tests were also frequent.

Nathan was finally diagnosed at five years of age when we visited a neurologist who instantly said the words "Sotos Syndrome" as we walked in the door. A bone X-ray was taken to determine bone age and a CAT scan was done to eliminate other causes of his symptoms. His growth charts were also looked at which concluded the diagnosis as being correct. At the same time his eyes were checked and he was found to be very short sighted with stigmatisms in both eyes.

What a relief it was to be told there was a name for Nathan's condition and a lot of questions were answered with the diagnosis. Still many other questions were unable to be answered as very little was known about the syndrome.

Nathan is now six years of age and is in his second year of schooling . He is in a special class in a mainstream school. We felt that due to safety aspects and the attention he would receive that this was the best environment for Nathan. So far he has been doing very well and feel that his school placement was the correct one for now.

The tantrums of the last few years are finally subsiding and so are a lot of his fears. He is generally a demanding but happy child and has the drive to attack life full on.

Nathan's future looks much brighter now than it did a few years ago and hopefully he may be able to become fairly independent later in his life. A life of his own is not out of the question.

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CONTACT CORNER

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

Anyone who wishes to reply to a request or a letter should write direct to the individual or group concerned where an address is provided. The AGSA office may be contacted for the information to be passed on in the case of anonymous requests,that is, where a name or address is not published please contact AGSA for details. Privacy and anonymity will be ensured if requested.

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

It should be remembered that a shared genetic condition does not mean an equally shared value system between families. Different degrees of acceptance and different mechanisms for coping will be encountered and a non-judgmental approach is recommended in establishing contact.

Goldenhar Syndrome

Alison would like to start a support group for Goldenhar initially by holding a coffee morning. Alison already knows of three other families and is looking forward to sharing experiences and ideas. Alison has a one year old daughter with this condition.

For further details contact:
Alison McAviney
23 Thompson Avenue
Illawong 2234
Phone 541 0389

Pseudoxanthoma Elasticum (PXE)

A family wishes to have contact with other families.

Acid Maltase Deficiency

Helen would like to meet up with another adult with this condition and she can be contacted at the following address:-

Helen Walker
2/1 Parnee Street
Swan Hill 3585
Phone 050 324093

Blephanophimosis Epicanthus Inversus and Ptosis Syndrome (BPES)

The parents of Christopher, aged two years and six months, would like to make contact with another family

Hydranencephaly - Schizencephaly

A family is seeking contact with another family with this condition.

Schwachman or Diamond Syndrome

A woman with a two year six month old son would like contact with another family.

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SUPPORT GROUPS

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.

Rare Chromosome Disorders Group Picnic Day

Peckys Playground Prospect played host for six families at the inaugural picnic for the Rare Chromosome Disorders Support Group.

After almost two weeks of non-stop rain in Sydney the morning of 12th March broke to cloudless skies and perfect picnic weather. Great distances were travelled with families from ACT, rural NSW, and the Central Coast attending. Although several apologies were received the attendance met expectations.

Disorders on Chromosomes 5,14, and 18 were represented , and an exchange of information on early intervention, respite care, schooling, etc occurred. This was done in an informal social atmosphere typical of picnics.

The need for the support group was reconfirmed by all in attendence. In view of the success of the day it was agreed that the group would meet again at a date to be fixed this coming September or October.

Our thanks must go to the staff at Peckys Playground which boasts outstanding facilities, immaculate grounds and a perfect venue.

Marlene and Scott Brightwell

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CONFERENCES

Osteogenesis Imperfecta
The Osteogenesis Imperfecta Association of Victoria is holding a conference on Saturday 13th May 1995, at the
Ella Latham Lecture Theatre
1st Floor of the old Building
Royal Children's Hospital
Flemington Road, Parkville, Victoria.

Contact:
Margaret Sandman Tel (03) 3065585
or

Margaret Sahhar, Social Worker, Murdoch Institute Tel (03) 345 5757

Alzheimer's
The 5th National Conference of the Alzheimer's Association of Australia will be held in Brisbane on 7-10 May 1995.

Contact:
Alzheimer's Association of Australia
PO Box 1280
Milton QLD 4064
Tel (07) 3690477
Fax (07) 3691512

"Tourette Towards 2000" Conference
To be held on 26-27 May 1995;
At the Park Royal Hotel, Darling Harbour
Cost: $10 per person per day
Telephone: 311 2745

For details of the following conferences which are all to be held in the USA , please contact the AGSA office.

Thalassemia Action Group 10th Anniversary Patient/Parent Group
April 29-30, New York State, USA

National Tay-Sachs & Allied Diseases Ass'n Annual Conference
May 5-7, Brookline, MA, USA.

National MPS Society Mini-Conference
May 6-7, Raleigh, NC ,USA.

Cornelia de Lange Syndrome Foundation Internatinal Conference
June 29-July 2, Orlando, Florida, USA

National Foundation for Ectodermal Dysplasias Conference
July 7-9, Grand Rapids, Michigan, USA.

Huntington's Disease Society of America Annual Convention
July 28-30, Philadelphia, PA,USA.

Battens Disease Support and Research Ass'n National Conference
August 3-6, Fife, Washington, USA.

Ehlers-Danlos National Foundation's National Learning Conference
August 15-18, Cincinnati, OH, USA

THE ASSOCIATION OF GENETIC SUPPORT
OF AUSTRALASIA INC.
FUNDED BY THE NSW HEALTH DEPARTMENT
REG. CHARITY No. C.C. 27702
66 Albion Street, SURRY HILLS NSW 2010
Tel: 211 1462 Office Hours:10-2 Mon-Fri

You are invited to the

LIVING GRIEF SEMINAR

Saturday 29 JULY 1995
Time: 10am to 5pm

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

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

The seminar speaker will be Dr John Rogers

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

Outline of the day

Confidentiality will be a key element of this Seminar

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

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

RSVP 1 JULY 1995

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

Payment Included; Yes ☐ **No** ☐

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

66 Albion Street
SURRY HILLS
NSW 2010

Tel: (02) 211 1462

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

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

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

ANNUAL SUBSCRIPTION

Individual	$15.00
Group/Organisation	$30.00

Subscription Year
1st October - 30th September

AGSA aims to:

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

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

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

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

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

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

HAEMOCHROMATOSIS INFORMATION SERVICE & SUPPORT GROUP
(IRON OVERLOAD)

NOTICE OF A MEETING.

Please take note of the following meeting to be held in SYDNEY.

DATE:- Sunday 20th August, 1995. **TIME:- 1:00pm to 4:00pm**

VENUE:-A.G.S.A. rooms, 66 Albion Street, Surry Hills, SYDNEY.

GUEST SPEAKERS:- Dr Mark Bassett MD FRACP (ACT) .. Margaret Rankin RN (QLD)
(Other guest speakers are currently being approached).

CONTACT FOR FURTHER INFORMATION:-

Lance McMillan
20 Neville Street
ST MARYS 2760

Tel:-(02)833-3216

Margaret Rankin
412 Musgrave Rd
COOPERS PLAINS 4108

Tel:-(07)345-7583

Dianne Petrie
c/- AGSA
66 Albion Street
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(02)211-1462