Contents

THE ASSOCIATION OF GENETIC SUPPORT OF AUSTRALASIA INC.

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

The UK Parliament announced approval for the utilization of Huntington Disease genetic test results in life insurance determination in Great Britain (www.doh.gov.uk/genetics/gaic.htm)

Reg Charity CFN154481 ABN 83 594 113 193

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Monday - Thursday
Closed Friday

ANNUAL SUBSCRIPTION

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Group/Organisation	$44.00

Subscription Year lst July - 30th June

PEER SUPPORT/INFORMATION OFFICER'S REPORT

The Olympic City is still glowing from the aftermath of the Sydney Games. One could not resist being moved by the Games. The Paralympic Opening and Closing Ceremonies were sold out for the first time in history and a record attendance was achieved at the sporting events. It was a special time in Sydney's history that will not be forgotten for a long time.

AGSA's AGM was held on 15th October and we are grateful to our guest speaker, Glenn Fisher who talked on her experiences as a volunteer at the Olympic/Paralympic Games. It is amazing how many of you who attended the Olympic Opening Ceremony saw Glenn Fisher on the big screen in the stadium talking about her experiences as a volunteer. Thank you Glenn.

I would like to welcome the in-coming committee members - Lance McMillan, Roberta Perla, Anne Mulder, Melanie Cameron and Scott Brightwell. A big welcome back to Richard Petrie who has been on the committee since 1988. Look for their profiles in the December Issue. We look forward to an exciting year ahead. Hopefully a year of securing more funding.

Best wishes

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

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Chromosome 13 q deletion syndrome

A family would like contact with others.

Thanatophoric Dwarfism

AGSA is looking for another family with this condition. Please contact AGSA for details

Sandoff Disease

A family in New Zealand is seeking contact with others.

Pseudo Pseudohypoparathyroidism

A lady would like contact with others to exchange management information.

ACT – Muscular Dystrophy Association Inc.

P O Box 2800, Tuggeranong ACT 2900

33 Allchin Circuit, Kambah ACT 2902

Telephone 02 62296 3000 Fax 02 6296 3901

National Toll Free Line 1800 010 040

NORTH QLD SHORT STATURED SUPPORT GROUP ESTABLISHED

My name is Heather Knuth, I live in Charters Towers with my husband Shane. We have a nine year-old daughter Anna, seven year-old boy, Joe, and a three-year-old Daniel. Daniel is short statured! At my 22-week scan they told me he had Down syndrome or would be a dwarf. When he was born, Achondroplasia was confirmed. I knew of no other short statured families, I was given no information and we felt we were alone. I contacted SSPA support group and got a bit of information about get togethers in NSW and Victoria but I wanted contacts in North Queensland, a bit closer. I thought surely there are others out there! When Daniel was two years old my paediatrician in Townsville kindly put me in touch with two mothers in Townsville who had one year old with SED. A friend put me in touch with another SED contact in Ravenshore (Atherton). We had a get together in Townsville and really enjoyed each other's support.

When Daniel was three years old, my paediatrician rang again; a one-month-old girl had been born in Townsville with Achondroplasia the same as Daniel. I drove down that week and met them with my photos and information. It was a beautiful morning, full of hope for their futures.

Then I had an article printed in Take 5 magazine about Daniel's story and a mother of an eleven-month-old boy with Heterozygous Achondroplasia contacted me from Mackay. She didn't know where to begin looking for someone in a similar situation and was eager to meet. We organized a meeting in Townsville and the local newspapers did a story. Our local Channel 9 News agreed to do a story and ten families turned up. A 74-year-old short statured man rang me after reading the article in the paper and was so glad to meet all the younger children.

Other contacts from Cairns came and we now have four contacts in Rockhampton area, so our next get together is for whoever can make it.

Once a term the Townsville folk try to meet in the school holidays. At our first official meeting in Townsville recently we had a birthday party for

three of the young ones turning 1, 2 and 3 year olds within a couple of weeks of each other. We had a

BBQ and all the children and parents got along like one big happy family. No one wanted to go home. It was comforting for the mothers to share stories and information and make new friendships. The fathers had a few drinks and got to know each other and the children made new lifetime friendships.

We will continue to meet whenever possible and are looking forward to the next one. If you know any short statured folk in North Queensland please ask them to contact

Heather Knuth
Coordinator
Nth Queensland Short Statured Support Group
P O Box 1351
Charters Towers Qld 4820
Ph 07 47878078

Thank you!

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

The following information is kindly supplied by the NSW Genetics Education Program.

COFFIN-SIRIS SYNDROME

Also known as

Dwarfism-Onychodysplasia

Fifth Digit Syndrome Mental Retardation with Hypoplastic 5th Fingernails and Toenails

Short Stature-Onychodysplasia

General information about Coffin-Siris syndrome

Coffin-Siris syndrome is a rare genetic or inherited disorder that may be evident at birth *(congenital)*. The features of this disorder may include:

- feeding difficulties
- frequent respiratory infections during infancy;
- diminished muscle tone *(hypotonia)*
- abnormal looseness *(Laxity)* of the joints
- delayed bone age;
- mental retardation
- short fifth fingers and toes with underdeveloped *(hypoplastic)* or absent nails

- other malformations of the fingers and toes.
- Characteristic abnormalities of the head and facial *(craniofacial)* area, resulting in a coarse facial appearance.

Craniofacial malformations may include

- An abnormally small head *(microcephaly)*
- A wide nose with a low nasal bridge
- A wide mouth with thick, prominent lips
- Thick eyebrows and eyelashes *(hypertrichosis)*
- Sparse scalp hair

The underlying cause of Coffin-Siris syndrome is unknown. In most cases, the disorder is thought to result from new genetic changes *(mutations)* that appear to occur randomly for unknown reasons *(sporadically)*.

Familial cases (i.e. more than one affected family member) have also been reported that suggest the disorder may follow one of two patterns of genetic inheritance in families – either autosomal dominant or autosomal recessive genetic inheritance.

Symptoms of Coffin-Siris syndrome in more detail:

Early in life, infants with Coffin-Siris syndrome typically experience feeding difficulties, vomiting, slow growth, and frequent respiratory infections. In addition, affected infants and children may have low muscle tone *(hypotonia)* abnormally loose joints, delayed bone age, and mild to severe mental retardation.

Individuals with Coffin-Siris syndrome also have characteristic skeletal or bony abnormalities. For example, certain fingers and toes particularly the fifth fingers and toes, may be unusually short due to absence or underdevelopment *(hypoplasia)* of the end bones *(terminal phalanges)* within these digits. The fingernails and toenails may also be underdeveloped or absent. Additional abnormalities may include dislocation of the inner forearm bone *(radius)* at the elbow, deformity of the hip *(coxa valga), or unusually small or absent knee caps (patellae)*.

Less commonly, affected individuals may have additional physical abnormalities, such as choanal atresia, a malformation in which a bony or thin layer of tissue blocks the passageway between the nose and throat, leading to difficulties breathing. Some individuals with Coffin-Siris syndrome may also have heart abnormalities at birth *(congenital heart defects)*. These may include persistence of the fetal channel that joins the main artery of the body *(aorta)* and the pulmonary artery *(patent ductus arteriosus)* or an abnormal opening in the fibrous partition *(septum)* that divides the upper or lower chambers of the heart *(atrial or ventricular septal defects)*. In addition, a brain abnormality known as Dandy-Walker malformation has been reported in some cases. This condition is characterized by cystic malformation and expansion of one of the cavities in the brain *(fourth ventricle)* Dandy-Walker malformation is usually associated with an abnormal accumulation of cerebrospinal fluid (CSF) in the skull *(hydrocephalus)*, resulting in increased fluid pressure, a rapid increase in head size abnormal prominence of the back regions of the head *(occiputal)* and/or other associated findings. Some individuals with Coffin-Siris syndrome may also have partial or complete absence of the band of nerve fibres that join the two hemispheres of the brain *(agenesis of the corpus callosum)*.

What causes Coffin-Siris syndrome?

The specific underlying cause of Coffin-Siris syndrome is not known. In most affected individuals the disorder is thought to result from new genetic changes *(mutations)* that appear to occur randomly for unknown reasons *(sporadically)*. In addition, some familial cases or more than one affected family member, have been reported. Some researchers suggest that such cases may represent a pattern of inheritance called autosomal dominant

Inheritance with variable expression i.e. symptoms will vary from patient to patient in both number and severity, even within the one family.

In some dominant disorders, potentially including Coffin-Siris syndrome, disease expression may be variable. In other words, if individuals inherit a mutated gene for the disease, the characteristics that are manifested may vary greatly in range and severity from case to case.

Other researchers indicate that Coffin-Siris syndrome may follow a pattern of genetic

inheritance in families called autosomal recessive inheritance.

Parents of some individuals with Coffin-Siris syndrome have been closely related by blood *(consanguineous)*. In recessive disorders, if both parents carry the same gene for the same disease trait, there is an increased risk that their children may inherit the two genes necessary for development of the disease. The chances of this occurring are greater when parents are related.

Some researchers also suggest that isolated *(sporadic)* and familial cases of Coffin-Siris syndrome may be due to unknown chromosomal abnormalities. Further research is required to determine the disorders underlying cause and potential mode of transmission.

Who is affected by Coffin-Siris syndrome?

Coffin-Siris syndrome appears to affect females about four times more frequently than males. Since the disorder was originally described in 1970 (G.S.Coffin), approximately 40 cases have been reported.

Is there any treatment for Coffin-Siris syndrome?

Diagnosis

It is possible that a diagnosis of Coffin-Siris syndrome may be suggested before birth *(prenatally)* based upon specialized tests such as ultrasound. During fetal ultrasonography, reflected sound waves are used to generate an image of the developing fetus. Ultrasound studies may reveal characteristic findings that may be associated with the disorder.

Treatment

The treatment of Coffin-Siris syndrome is directed toward the specific symptoms that are apparent in each individual. Such treatment may require the coordinated efforts of a team of medical professionals who may need to systematically and comprehensively plan an affected child's treatment.

In some affected individuals, treatment may include surgical repair of certain craniofacial skeletal, cardiac, or other abnormalities potentially associated with the disorder. The surgical procedures performed will depend upon the severity of the anatomical abnormalities, their associated symptoms, and other factors.

In addition, in those with choanal atresia, surgery or other appropriate methods may be required to decrease the airway obstruction or correct the malformation. If affected individuals have Dandy-Walker malformation, treatment may include surgical implantation of a specialized device *(shunt)* to drain excess cerebrospinal fluid (CSF) away from the brain and into another part of the body where the CSF can be absorbed. During infancy, treatment may also require measures to help prevent or aggressively treat respiratory infections.

Early intervention may be important in ensuring that affected children reach their potential.

Genetic counselling is recommended for individuals with Coffin-Siris syndrome and their families.

FAMILY STORY

ALEXANDRIA CUMBERLAND

United Kingdom

DOB. 23-7-97. **Weight** 2.7kg.
Length 49cm.

Alex, our second child was born at 39 weeks gestation, following a normal pregnancy and delivery. Her apgar scores were 9-9-9, she appeared to be ok. From the very first attempted feed, she didn't seem interested. [Or so we thought]. Five days of trying to get her to breastfeed, to no avail. Bottle-feeding was just as hard, in the end we just had to squeeze the formula into her.

Whilst still in hospital, she had the usual checks by the Paediatric Doctor. We were asked about certain

features that Alex had. We later found out i.e. 20 months later what had been written, on her discharge sheet. We had never heard of these words, dysmorphic features before. We will never forget them now.

At 10 days old Alex was seen by our health visitor who noticed an unusual cry, called a stridor. Alex seemed to be a good baby, very quiet and slept a lot.

Alex was always a sickly baby right from the start. She could be sick, right up until her next feed. We were given Infant Gaviscon to thicken up her milk feed. It never worked.

At first we thought it was an allergy to milk, then eventually we found out by our own process of elimination. She has allergies to apple produce and gluten products. Our paediatrician will not test her for coeliac disease yet!

We started to notice from the age of 4-6 months that she wasn't doing things i.e. smiling and reacting to noise. At 10.5 months she was just about sitting up, we requested to see a paediatrician, regarding her development delay. At 14 months she was seen by the community paediatrician and we were told that she had a developmental delay of 8 months. And that her speech as at the age of 6 months. From there she was referred to the Child Development Centre.

At the same time, we were told that she had moderate sensory hearing loss. So from then on, she wore 2 hearing aids. She wasn't impressed with the idea! She wore hearing aids until she had grommets inserted in Nov 99. She doesn't have Sensory Hearing loss; it was severe glue ear!

At first they blamed the hearing loss for her delay. Then she was referred to the Regional Genetics team based in Manchester. We saw them in July 1999 and on the 13th October we were given a diagnosis of Coffin Siris Syndrome. Our lives changed forever!

We were told that they had seen another girl with C.S.S. but they didn't know where she was. We have since had contact with the family.

Her milestones are sitting at 10.5 months, crawling at 18 months, walking at 2yrs 2 months.

Alex is a happy sociable child. She is beginning to enjoy interacting with her siblings, Jonathan [6] and Rebekah [20months]. At home she loves to play in the garden, watch TV, look at books. She is very inquisitive and loves climbing!

Her speech and language are very delayed, but she has been assessed as having some cognitive and social foundation skills. She understands 10 Makaton signs. She copies the drink and yes sign, when prompted by an adult.

Alex has scoliosis. When first x-rayed, her spine was found to have twist of 30 degrees. That was back in January 2000. Now her spine has a twist of 40 degrees. Her orthopaedic surgeon has told us she may need surgery when she has stopped growing, as she is a very active child, he will not fit her with a brace. He sees no actual benefit for her.

She attends a special needs nursery school, which she loves very much. We have noticed a big change in Alex's development, since she has been attending Broadlands. It is a very slow process, but she'll get there at her own pace.

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TAXI SUBSIDY SCHEME

Australia has a National Taxi Subsidy Scheme that ensures eligible people with disabilities who are members of State Subsidy Schemes can receive their home state entitlements when travelling interstate.
The Taxi Transport Subsidy Scheme
can be contacted on 02 9270 6080
Freecall 1800 623 724 for more information

CONFERENCES

Provisional Development Course

Challenging Behaviours

27th, 28th, 29th, 30th November &

lst December 2000

Cost $100 Contact Special Education & Disability Services (08) 8288 0827

Managing Problem Behaviours in People with Intellectual Disabilities and Autism Spectrum Disorders on

Friday 10th November 2000

Venue: Stephen Roberts Theatre
The University of Sydney

International Keynote Speakers
Professor Ian Wallander
Civitan Center
The University of Alabama, USA

Cost $150.00 Contact Mrs Helen Moore

Ph 02 8878 0500

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Paediatric & Child Health Nurses 2001 Conference

17-19 May 2001

Carlton Crest Hotel, Brisbane, Qld.

Call for Abstracts
Contact
Secretariat
P O Box 1280
Milton Qld 4064
Tel +61 7 3858 5561

The Social Relations of Disability Research Network with People with Disabilities (NSW) Women with Disabilities Australia and Access Plus
invite you
to participate in an International Conference.

DISABILITY WITH ATTITUDE; CRITICAL ISSUES 20 YEARS AFTER IYDP

Keynote speaker confirmed
Dr Tom Shakespeare,
(University of Newcastle, UK)

Since International Year of the Disabled Person (IYDP) in 1981, people with disabilities have been pressuring governments to address discrimination and enhance their wider social participation, self-determination and personal autonomy. Issues of rights, recognition and resources have been central to these struggles. This conference offers a forum for assessing progress 20 years after IYDP, celebrating achievements and examining critical issues facing Australian society over the next two decades. It also provides an opportunity for participants to engage in a critical audit of Australia's record on disability issues, and to do so within an international framework. Major themes to be addressed involved identity, gender, participation, representation, sexuality, violence and abuse, and the relationship between the disability movement and academic scholarship.

Phone 02 9385 1879 for details.

An Australian Academy of Science Boden Research Conference on Immune Deviation and Reproductive Function

February 19-21, 2001

Erskine House, Lorne, Victoria.

Jointly sponsored by the Fertility Society of Australia, The Australasian Society for Immunology, and the Society for Reproductive biology.

The immune system is an integral component of the normal physiology for all reproductive tissues. This timely conference brings together prominent international and Australian scientists and clinicians working at the cutting-edge of this research. Symposia topics will cover the new immunobiology of the male and female reproductive tracts, immunological factors affecting implantation, the role of the immune system in reproductive dysfunction and sexually transmitted diseases, and the current status of contraceptive approaches using immunological approaches.

For more information
Contact
Mark Hedger, Monash Institute of Reproduction and Development
62 3 9594 7124

OVERSEAS CONFERENCES

8th World congress of Gynecological Endocrinology, Florence, Italy
6th-9th December 2000

Scientific Secretariat:
Dept., of Reproductive Medicine & Child Development
Div. of Gynecology and Obstetrics
"P. Fioretti"
University of Pisa, Via Roma 35,
56126 Pisa – Italy

Fax 0039-050-553410

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Endocrine Society of Australia

Annual Seminar Meeting for Physicians and Scientists, Newcastle. New South Wales

20th-22nd April 2000

David Maddison Building,
Newcastle
NSW

The theme for the first year of the three year programme will be Pituitary and Thyroid Pathology and Paediatric Endocrinology.

International Invited speakers:

Dr Sylvia Asa, Pathologist in Chief, University Health Network and Toronto Medical Laboratories Toronto Canada

Dr Bin Tean The, Senior Investigator, Van Andel Institute, Grand Rapids, Michigan USA.

Topics to be Covered

- Developmental genes in the pituitary
- Hypopituitarism
- Unusual tumours of the hypothalamus and pituitary including craniopharyngioma
- Role of radiotherapy
- Multiple Endocrine neoplasia – new genes and syndromes
- Familial acromegaly
- Developmental genes in the thyroid
- Controversies in Thyroid Pathology
- Thyroid tumours – pathogenesis and management
- Thyrotoxicosis in pregnancy
- Congenital Hypothyroidism

Convenor

Dr Patricia Crock
Dept of Paediatric Endocrinology,
John Hunter Children's Hospital,
Newcastle NSW 2310.
Ph: 02 4921 3672
Fax: 02 4921 3599
Email: pcrock@mail.newcastle.edu.au

Developmental Disability Clinic

This Clinic works in conjunction with the person's general practitioner, to ensure access to high quality health care through a network of allied health personnel.

Health needs of people with a Developmental Disability.

People with developmental disabilities often have health problems which are greater than those of the general community. Many of these needs can be met by a person's general practitioner, but sometimes a specialist approach is required.

For this reason the Centre for Developmental Disability Studies has sponsored a health clinic at the Royal Rehabilitation Centre Sydney. This clinic provides a consultant service for people with Developmental Disabilities and their carers.

The clinic works in conjunction with the person's medical practitioner, to ensure access to high quality health care through a network of specialists and allied health personnel.

If there are behavioural problems, the clinic will identify any possible physical cause, and refer for behavioural advice as required.

Contact
Royal Rehabilitation Centre Sydney
259 Morrison Rd Ryde NSW 2112
Phone 02 9808 928

SUPPORT GROUP

For Relatives and Friends of People with Tourette's syndrome "Plus"

Venue: Rotary Lodge, Reserve Road, Royal North Shore Hospital.

Date and Time: Fourth Wednesday of every month 6:30 p.m.– 8:30 p.m.

Contact Person: Jenny Papa 9427 1254

TS+

The aim of the group is to be mutually supportive through shared experiences and professionally facilitated discussion groups. To learn from each other and guest speakers, and to foster Tourette's syndrome education in government and the community by active participation of members.

'+' Stands for other disorders that tend to be comorbid with TS such as; Attention Deficit Hyperactivity Disorder (ADHD), Attention Deficit Disorder (ADD), Obsessive - Compulsive Disorder (OCD), Panic attacks, Anxiety, 'Rage', Depression and Specific Learning Disabilities.

AGSA SUPPORT GROUP MEMBERS as at February 2000

Act Muscular Dystrophy Association Inc.
Androgen Insensitivity Assoc.Support Group of Australia
Alagille Syndrome Support Group
Albino Support Group
Alliance of Genetic Support Group, U.S.A.
Angelman Syndrome Assoc. Inc.
Alzheimer's Assoc of Aust Inc.
A.P.I.A. (Aust.Primary Immune Deficiencies Assoc.)
Assoc. for Children With a Disability, Vic.
Assoc. for the Welfare of Child Health (AWCH)
AUSSIE FOLKS
Aust. Arthrogryposis Group (TAAG) Inc.
Aust. CHARGE Association
Aust. Crohn's & Colitis Assoc.
Aust. Huntington's Disease Association (Qld) Inc.
Aust. Huntington's Disease Assoc. (NSW) Inc.
Aust. Speak East Assoc.
Australasian Tuberous Sclerosis Society Inc.
Aust. Leukodystrophy Support Group
Aust. Society for Ectodermal Dysplasia
Autistic Assoc. of NSW
Batten's Disease Support & Research Foundation
Beckwith-Weidemann Syndrome Support Group
Cardiomyopathy Assoc of Aust. Inc
Centacare Early Intervention.
Charcot Marie-Tooth Assoc. of Aust Inc.
Charcot Marie-Tooth Disease, USA
CONTACT A FAMILY U.K.
Cleft Pals, The Cleft Palate & Lip Society
*CLIMB **C**hildren **L**iving with **I**nherited **M**etabolic **D**iseases*
Coeliac Society of NSW Inc.
Congenital Adrenal Hyperplasia Support Group
Cornelia de Lange Syndrome Support Group
Cri du Chat Syndrome Support group of Australia
CVS Support Group (WA)
Cystic Fibrosis Assoc of Qld Ltd.
Cystic Fibrosis Assoc. of Vic
Cystic Fibrosis Foundation, North Ryde.
Early Education Clinic, North Sydney
DIAL (Qld)
Donor Conception Support Group
Depressive & Manic Depressive Assoc.
Dystrophic Epidermolysis Bullosa Research Association (DEBRA) NSW. Inc.
Ehlers-Danlos Syndrome Support Group
Exceptional Parent (USA)
Fabry's Support Group Inc.(Vic)
Family Advocacy
Family Planning Assoc.
FAP Register (NSW Cancer Council)
Fragile X Assoc of Australia
Friedreich Ataxia Assoc of NSW
Gaucher Assoc. of Australia
Genzyme Australia Pty. Ltd.
Genetic Interest Group (GIG) (UK)
I.D.E.A.S. Inc
Klinefelter Syndrome Support Group
Kurrajong Early Intervention
Haemochromatosis Information Service & Support Group NSW
Haemophilia Foundation NSW
Hereditary Haemorrhagic Telangiectasia
Hereditary Fructose Intolerance
Hunter Orthopaedia School
Huntingtons Disease Assoc. (NSW)
Huntingtons Disease Assoc. (QLD)
IDEAS Onc.
Maternity Alliance
NALAG
Leukodystrophy Foundation (USA)
Leighs Disease Support Group
Lowe's Syndrome Assoc. Inc.(USA)
Lower Nth Shore Community Support Team
Lysosomal Diseases Australia
M.P.S. Society
Marfan Syndrome Support Assoc. NSW
Marfan Syndrome Assoc.Australia (S.A.Branch))
Meniere's (NSW) Support Group
Motor Neurone Disease Assoc. of NSW Inc.
Multiple Epiphyseal Dysplasia Assoc.
Muscular Dystrophy Assoc of NSW
Muscular Dystrophy Assoc (NZ) Inc.
National Council of Intellectual Disability
Neurofibromatosis Assoc.
Noonan Syndrome Support Group
NSW Genetics Education Program
NSW Cancer Council
Osteopetrosis Support Group
Osteogenesis Imperfecta of Aust.
Parents Bereavement Support Group
Parent to Parent (NZ)
Pen-Parents of Aust. (ACT)
PKU Assoc of NSW
Psoriasis Society
Pseudohypoparathyroidism Support Group
Pseudoxanthoma Elasticum Support Group
Prader-Willi Syndrome Assoc
Pyruvate dehydrogenase deficiency.
Rare Chromosomes Disorders Support Group
Retinitis Pigmentosa Society of NSW Inc.
Rett Syndrome Assoc. of Aust.
Royal Blind Society of NSW
SAFDA (Support After Foetal Diagnosis of Abnormality)
SANDS
Short Statured People of Northern Qld
Short Statured People of Aust (NSW)
Short Statured People of Aust (Vic)
Short Statured People of Aust. (SA)
Spinal Muscular Atrophy
Schizophrenia Fellowship NZ
Smith Magenis Syndrome Support Group Inc.
Spastic Society of Victoria
Spina Bifida Assoc. of NSW
Spina Bifida Assoc. of WA Inc.
Society of Ectodermal Dysplasia
SOFT Australia
Southern Child Care Support Program
Sotos Syndrome Support Group
The Chromosome 18 Registry & Research Society
The Northcott Society
Thalassaemia Society of NSW
Turner Syndrome Assoc of Aust. Ltd. (QLD)
Turner Syndrome Assoc. of Aust. Ltd. (NSW)
Uncontrolled Epilepsy Support Assoc (Vic)
United Leukodystrophy Foundation (USA)
Velo-Cardio-Facial Syndrome Foundation of Australia.
Wellington Huntington's Disease Assoc.(Inc.) (NZ)
West Syndrome Support Group
Wolf-Hirschhorn 4p- Syndrome Support Group
Williams Syndrome Association of Aust. Inc.

*(**NB**: This list represents support groups and associations only. In addition to this list of members AGSA has established a Contact Register over 500 genetic conditions representing families and individuals seeking contact.)*