



**THE ASSOCIATION
OF GENETIC SUPPORT OF
AUSTRALASIA INC.**

66 Albion Street, Surry Hills, NSW 2010
Australia
(02) 9211 1462

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NEWSLETTER

APRIL 1999 ISSUE 41

MISSION STATEMENT

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

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Contents

Contact Corner: Bannayan-Zonana syndrome,
Borgeson Forffman Lehman syndrome,
Pachymychia Congenita, Robinow Syndrome,
Ruvalcaaba-Myhre-Smith syndrome.

Profile: Sturge Weber syndrome
Siblings within the Family Unit where there is a
child with a Developmental Disability

**GENETIC DISORDERS
AWARENESS WEEK
16th -22nd June 1999**

Launch 16th June 1999

Guest Speaker: Dr Grant R Sutherland AC
*Director, Department of Cytogenetics and
Molecular Genetics, Women's and Children's
Hospital and Affiliate Professor in the
Departments of Paediatrics and Genetics of the
University of Adelaide. He is a Fellow of the
Royal Society of London and of the Australian
Academy of Science. He is a past-President of
the Human Genetics Society of Australasia and
of the Human Genome Organisation. His
research interests are the Human Genome
Project and the molecular genetics of mental
retardation and epilepsy. He was a co-recipient
of the 1998 Australia Prize for molecular
genetics.*

**Coles Theatre, Powerhouse Museum
Level 3 (entry via steps underneath Luna
Park Face)**

500 Harris Street, Ultimo NSW 2007

Wednesday, 16th June 1999 at 6.00 pm.

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

PEER SUPPORT/INFORMATION OFFICER'S REPORT

I write to you from the hailstorm damaged Eastern Suburbs. Each day I see further evidence of the April 11th storm. The roar of the storm approaching was frightening. Many in our street thought a plane had crashed or a tidal wave was about to hit. Hailstones the size of cricket balls smashed car windows, house windows, tiles, TV aerials, garden furniture and thousands of skylights causing extensive flooding. Nearly every house in the area with a tiled roof now has a tarpaulin covering the roof. The clean up will take up to 9 months and cost over \$1 billion. Imagine if that money was spent on genetic research and disability services.

At AGSA's recent Support Group Seminar it was agreed by the majority of people attending that it was good to get together to network and exchange ideas and they would like to see this continue but it was disappointing two speakers were unable to attend. The speaker from the South Sydney Council could not attend because the Council Chambers were flooded during the storm and the other speaker had hurt her ankle. Ross Johnson from the South East Sydney Area Health Service gave an interesting talk on grants available for non government organisations.

Genetic Disorders Awareness Week is fast approaching and I look forward to seeing you at the launch on 16th June at the Powerhouse Museum. It promises to be a great night. Genetic Disorders Week is a recognition of the wonderful work support group coordinators do and an acknowledgment of the partnerships forged between the genetics clinics. It is also an important time to create awareness of the unique support AGSA and support groups provide. AGSA is strongly supported this year by the Ageing and Disability Department and the Powerhouse Museum.

Until then, 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.

BANNAYAN-ZONANA SYNDROME

Please contact Kim Frumer, Genetic Counsellor, Port Macquarie, if you know of another family with this condition.

BORGESON FORFFMAN LEHMAN SYNDROME

Angela Stephenson, Genetic Counsellor, Tamworth, is looking for contact for this family. Please contact her if you know of another family.

PACHYOMYCHIA CONGENITA

A mother of a four and half year old baby boy would like contact with another family. Please contact the AGSA office for details.

ROBINOW SYNDROME OR FETAL FACE SYNDROME

A mother of two children with this condition would like contact with another family. Please contact AGSA for details.

RUVALCABA-MYHRE-SMITH SYNDROME

Please contact Kim Frumar, Genetic Counsellor, Port Macquarie, if you know of another family who would like contact.

The Equity and Tax (EaT) Campaign

As the Senate debate intensifies on the Howard Government's tax package, ACOSS has launched its Equity and Tax (EaT) campaign, with three pamphlets. The campaign seeks the support of the community sector and the general public to:

- * Keep all food GST-free;
- * Shave the income tax cuts and close more loopholes; and
- * Stop the introduction of the GST or tax cuts before key business tax changes are legislated.

The pamphlets note, in relation to the GST, that "No government can ensure that everyone is compensated and no government can commit future governments to maintain this compensation."

Copies of the EaT pamphlets are available from ACOSS on (02) 9310 4844.

FAMILY STORY

Reprinted with the kind permission of The Weekend Australian 27-28 March 1999.

Syndrome has family searching

by Belinda Hickman (Medical Writer)

Jacinta Stanford was born two years ago with a crimson birthmark covering the right side of her head.

Doctors thought the mark - called a port-wine stain - would turn out to be harmless and told her parents, Joanne and Peter, it could be removed eventually by laser.

But by the time Jacinta was four months old she had begun to have seizures, and it became apparent the birthmark was a sign of more extensive and complex problems.

Jacinta was diagnosed with Sturge Weber syndrome, a rare neurological condition involving excessive growth of blood vessels.

The port-wine stain is caused by blood vessels growing under the skin. Neurological symptoms are caused by an underlying mass of vessels covering the surface of the brain.

The mass constricts oxygen flow to the tissue underneath, limited the brain's growth and development and eventually causing it to shrink. In Jacinta's case, symptoms are limited to one half of her face, but for some children excessive blood vessels can grow on both sides of the head. In a related condition, the growth occurs not on the head, but in the limbs or on internal organs.

Ms Stanford says the diagnosis has set the family on a path of discovery via the Internet and e-mail as there is little information available in Australia about the condition and few doctors have come into contact with it.

"You start off thinking it is just a birthmark and then you hear about all these other things that could, and have been, going wrong," Ms Stanford

says. The couple obtained most of their information from the US based Sturge-Weber Foundation which funds most of the research being done into the disorder. Ms Stanford has become its Australian representative. The incidence of the rare syndrome is difficult to pinpoint, but Ms Stanford knows of 28 Australian families with affected children who have joined the foundation. In July, the couple will travel to the US at their own expense to attend the foundation's bi-annual conference for families, researchers and specialists. They hope to return with information to help Jacinta and other children with the disorder progress.

Already Sturge-Weber syndrome has cast a shadow over Jacinta's life, affecting her health and development.

By nine months of age the infant was having seizures every day, sometimes lasting one or two hours. As a result, she developed only to the level of a four-month old child.

"She couldn't learn to do anything because she'd keep getting bombarded with the seizures," Ms Stanford says.

The infant has laser treatment every three months to help fade the birthmark on her skin, and last year she underwent delicate surgery in which doctors removed the damaged half of her brain.

Jacinta is one of 75 children aged up to 15 years who, over the past decade, have had the surgery performed by doctors at the Children's Neuroscience Centre at Melbourne's Royal Children's Hospital.

The director of the hospital's neurosurgery department, Prof Jeffrey Rosenfeld, says the operation involves doctors removing dead and damaged brain tissue, which impairs a child's development.

Since the operation, Jacinta has improved markedly, her mum says.

"Within weeks of the operation we could see the difference in her face, in the way she looked. There was a clarity in her eyes".

Jacinta has had only two seizure episodes in the past year, has mastered the art of scooting around on her bottom and is learning to walk. While she is likely to remain weak on one side and may never use her left, hand, it is too early to tell whether the seizures have caused lasting intellectual disabilities.

Her parents are hopeful.

* Information is available on the Sturge-Weber Foundations' Web site at:

<http://www.sturge-weber.com>.

SIBLINGS WITHIN THE FAMILY UNIT WHERE THERE IS A CHILD WITH A DEVELOPMENTAL DISABILITY

by **Margaret Cupitt**

Every family member is affected in some way when a child is born or diagnosed as having some form of developmental disability but actual impact will vary from family to family and individual to individual within each family. It's not possible to concentrate solely on siblings without some consideration of the affects on the whole family.

If we consider the normal stages of development, it can be seen that the environment in which we grow is one of the biggest influencing factors especially in the early formative years. Our initial attitudes, values, the way we interact with others - in fact act, most of our social skills, are based largely upon those of our parents and our immediate and extended family members. As we grow and start to interact with a wider range of people at school and in out of school activities we are often confronted by different values and attitudes. Eventually we develop our own set of values and attitudes based on a mixture of family and society at large.

Siblings relationships critical to a child's overall development take on special significance when

one of the siblings has a disability. It is difficult to understand completely why the sibling relations alters when a child has a disability. There are a multitude of factors involved and the individual nature of each family make it impossible to clarify the uniqueness and specialness of these sibling relationships.

Following is an excerpt from Margaret's article on how siblings view themselves.

THEMSELVES

Many children ask the question "Will I catch the disability?" One child in a sibling program had been told that her sister's disability was caused because mum had diabetes and this little girl, who was 9 years of age at the time, was quite sure that sooner or later she would catch this disability as well. She couldn't understand that if she'd grown in her mother's tummy just as her sibling had, why, then, wasn't she also disabled.

Children do ask the question "Is there something wrong with me as well?" They see that their sibling has a disability then they also think there must be something wrong with them, even if it's not the same sort of disability.

Relationship with Siblings

Questions are asked like "How can I get along better with my sibling? Will we have a normal brother/sister relationship?" This last point is particularly reinforced when they start going to school and developing friendships with their peers and seeing the sort of interaction there is between their peers with their brothers and sisters. Children ask "Why am I smarter, stronger than my sibling? How come he/she is the one with the disability? How come it wasn't me?" They often feel very guilty and perhaps feel they should perhaps be more loving, and behave better with their brother or sister.

Participating in Child Rearing

At quite a young age, children often feel that they would like to express their views on raising siblings, it becomes more evident as the children get older. In the adolescent and young adult it often seems that there are things that parents are

doing with their sibling that they disagree with, but are not sure whether they should express their views or not. Again, as siblings are getting older, they can sometimes be very resentful if they're always being asked to babysit, feeling responsible, whether that be at home, when they're out or at school.

In some situations where the child with a disability is attending the same school as their sibling, the siblings are called if the child with the disability needs any attention or if a situation arises, then the sibling is often called to come and assist. The children feel great responsibility regarding the care for their sibling.

Themselves and their Feelings

'Why do I have such mixed feelings?' "Why do I have this love/hate relationship?" - and yet to look at the interactions and relationship between siblings generally, most people can think of times when they have had a love/hate relationship with their own sibling. So because one child has a disability it doesn't necessarily mean that that's why there is a love/hate relationship developing. However, the difference probably lies where one child has a disability, and the sibling tends to feel a little bit more guilty if he/she gets angry or if he/she has what we would term "negative feelings" towards the sibling. It doesn't seem to have the same impact when there are two siblings without disability.

FRIENDS

Informing Friends

As children develop relationships with peers, there are a lot of issues that can come to the fore, such as "How or what is the best way to tell my friends about my sibling?" or wondering whether, in fact, they should say anything. They can be sometimes very self conscious and embarrassed when they first invite a friend home because they are concerned as to whether their friends will really understand. They may wonder whether, when their friends find that they have a brother or a sister with a disability, the friends will think that they are disabled too. "Perhaps my friends will think that they can catch the

disability". There is another big issue for them - the concern that perhaps the friends may go back and tell everybody at school. The embarrassment of having everybody else in their class or others within the school knowing something that they see as private, may be distressing and intrusive.

Their Friends' Acceptance

When children do bring friends home, it's often difficult for the siblings to know whether or not they should invite their brother or sister to play with them. They worry about the sort of reception from their friends - whether the friends will accept him or her, whether they'll like him or her or whether they'll be afraid of their brother or sister.

Teasing

It's important to put this in the perspective of peer pressure. It is unfortunate that children can be so very cruel and when teasing and name-calling starts, often other kids are labelled as being chicken or some other name that may be even more derogatory if they don't participate. And so children who have a brother or sister with a disability are faced with this on a far more personal basis. "What should I do when my friends tease my sibling?" and the conflict of feeling at times some peer pressure to join in the teasing but at the same time feeling guilty about that and thinking "Why are others so mean to my sibling?" It can be a double standard: "It's OK for me to do something to my brother or sister but it's not OK for somebody else". The dilemma lies in really not knowing what to say to friends or to other children about the same age who are participating in this teasing and inappropriate behaviour.

Often children will torment the non-disabled child and kids can feel quite angry and upset about the fact that they are being teased about their brother or sister.

Another question arising is "What should I do when kids make fun of other people who have a disability?" and they will see this as in some way reflecting back on their own brother or sister.

THE COMMUNITY

School

If a child is attending a special school or a special support class within a regular school, often other family members are unsure of what the special class or what the special school really is. It is therefore important that the other siblings are given an opportunity to visit their sibling's school and become acquainted with the environment that their brother or sister is involved in. They are generally interested in what their sibling is learning and whether the teachers treat the children in the special class or at the special school the same as they're treated in their own school.

One of the big concerns for children who have a sibling attending a regular school is that other kids will be cruel to their sibling. In a program that was run in early 1989, there were children who were very adamant about their feelings that their brother or sister should not be attending the local regular school. The reason for this was that the other kids were too cruel and they didn't want to see their brother or sister treated like they'd seen some other children with a disability treated. The non-disabled child finds that they're being compared with their sibling either in terms of things that they can do, or looking to see whether they too may have a disability. Again, the concern of being responsible for the sibling's behaviour, and where this has been a problem, that they will be called to assist with their brother or sister causes much embarrassment. They question "Why do teachers call me when there's a problem?" However, the reverse of that is that often they become really upset or angry when they're not called if there is a problem; and so again it can become a slight Catch-22 situation. Often the concern is "Will I have to associate with my sibling at school or can I pretend that I don't know who he or she is?"

Community acceptance

Again one of the issues arising is whether their sibling will be accepted in public. Parents feel embarrassed when a child is behaving inappropriately in public, similarly siblings

probably feel that embarrassment even more, questioning "Why do people stare at us when we're out?"

Another aspect of public acceptance is being unsure as to what to say to strangers. If strangers ask about the child with a disability, what should the response be? They're concerned that if their sibling has a seizure in public - "What should we do?" and again there is the embarrassment of perhaps feeling that attention has been drawn to them because of something that's happening.

There is a genuine concern that people will take advantage of the sibling with a disability and that they may be exploited by other people. Another issue for children is simply whether anyone outside the family will be a friend to the person with the disability.

ADULTHOOD AND GUARDIANSHIP

At quite an early age children become concerned as to "Who's going to take responsibility for my sibling when my parents die?" Although they may see that at age 8 they can't take care of their sibling, they may certainly be very aware of the fact that should something happen to mum and dad, then there is a great need for somebody who has the same care and love that the parents have to continue to look after the sibling. As they get older they may wonder whether, in fact, they will have to take that responsibility and the parenting role. "Will I be financially responsible?" "Will my sibling have to live with me?" and "If I'm not guardian and I'm not taking this care and responsibility for my sibling, then who will?"

Their own family

During the adolescent years, kids start to question what the long term affect of having a sibling with a disability will be: if they get married, "Will my spouse accept my sibling?" "What about my own children? If I have children, are they likely to have a disability as well?" The need, perhaps for genetic counselling and talking to somebody about these concerns is really important, looking at balancing their own needs against the perceived responsibilities towards their sibling - "Is it really going to be

necessary for my sibling to perhaps come and live with me, and what affect would that have?"

Continuing Involvement

Where one may continue to expect to have contact and involvement with siblings as we get older, it takes on different context when one of those siblings has a disability. The question often comes up: "How should I continue to be involved with my sibling?" especially if he or she is living in a group home. The need to look at the sort of role and responsibility that they may take on: whether they should visit the group home; bring their sibling home for weekends, whether they should advocate on his or her behalf, perhaps join a parent/sibling group. But perhaps the other side of that is the guilt that's attached if the sibling doesn't want to be involved and says "But I don't even want to see my sibling any more". When that happens in some families where there isn't a member with a disability, there is far less guilt than if one child has a disability.

Strategies for Parents.

Be open and honest.

Value each child's individuality and each individual child.

Limit care giving responsibilities.

Make good use of respite care and other support services.

Be fair when dealing with discipline, attention and resources.

Accept the disability.

Have books and articles available for children relating to disability.

Schedule special time with each sibling.

Welcome other children and friends into your home.

Praise siblings.

Involved the siblings in the life of the sibling with a disability - school meetings, going and visiting the school, therapy programs, assessments and visits to professionals.

Require the child with the disability to do as much as possible for him or herself.

Encourage participation of children in siblings groups.

Use professionals to help siblings.

Recognise special stress times for siblings. There are specific times that are often more stressful or sensitive times for children and it's just good to be aware of these times. they are not much different to perhaps the more vulnerable times for parents:

- * The birth of another child
- * The child with the disability goes to school
- * Sibling starts to date
- * Friends reject the child with the disability
- * Friends ask questions about the child with the disability.
- * The child with the disability becomes critically ill
- * If problems relating to the child with the disability are handled in secrecy
- * Parents become ill or die
- * When the siblings marry.
- * Provide opportunities for normal family activities.
- * Don't expect sibling to be saints.
- * Participate in activities involving the non-disabled child. It is important that the other children feel that you're interested in the things that they are doing, whether that be in social activities, soccer club, netball; their school activities - whatever it might be, but try to maintain the time.

Try to keep it all in perspective and Good Luck!

CONFERENCES

“Cancer: We Care”

National Conference

for Cancer patients, carers, volunteers and health professionals.

Key issues to be covered:

Survivorship/living with cancer; Treatment Advances; Recognising the carer; Advocacy and Communication Skills.

13 - 16 July 1999

Australian Jockey Club
Convention and Exhibition Centre
Randwick NSW

For more information contact

Ms Emma Mesaros, Conference Coordinator
Australian Cancer Society
GPO Box 4708
Sydney 2001
Ph: 02 9380 9022
Fax: 02 9380 9033
Email: acsapo@cancer.org.au

IDEAS EXPO 99

Albury Convention & Performing Arts centre
Swift Street, Albury NSW

Thursday 20 May 12 - 5 pm

Friday 21 May 9 am - 5 pm

Professional Seminar “Ageing with a Disability”

Saturday 22 May 9am - 1 pm.

Contact IDEAS Inc. 1800 029 904

Fax: (02) 69473 723

NCOSS HEALTH CONSUMER EVENT:

Health Financing Summit

Tuesday 25 May 1999

Furama Hotel, 28 Albion Street, Surry Hills,
Sydney

Giving consumers a voice in health policy.

Contact NCOSS 9211 2599

The New Children's Hospital

**"Challenging Behaviour in Individuals with
Developmental Disabilities"**

Saturday 19th June 1999

Lorimer Dods Lecture Theatre

The New Children's Hospital
Cnr Hawkesbury Rd & Hainsworth St
Westmead

9 am - 4.30 pm

Contact: Prof Jeff Sigafos

Ph: 02 9361 6112 Fax 02 9351 6469

**Physical Disability Council of NSW
1999**

**Consultations and Workshop
Seminars**

"Affecting the Agenda Sitting Down"

Dates & Locations:

Grafton - Fri 30 July *Training in lobbying*

Bankstown - Wed 11 August

Care and support and training in lobbying

Griffith - Mon 13 September

Training in lobbying

Chatswood - Sun 31 October

AGM - Citizenship

**7th National Conference
Association for the Welfare of Child
Health**

*"Through the looking glass
Lessons from the past for the future of
children's health care."*

7- 8 October 1999

Sydney Children's Hospital, Randwick NSW

Contact: (02) 9633 1988

Fax : (02) 9633 1180

**The Australasian Tuberous Sclerosis
Society Inc. Invites**

all members and interested professionals to
attend the

ANNUAL FAMILY CONFERENCE

to be held on the weekend of

14 and 15th August 1999

at the

Royal Institute for Deaf and Blind Children at
North Rocks, Sydney.

The conference will provide opportunities to
learn more about Tuberous Sclerosis from
experts and other families affected by this little
known disease. Meeting others who share T.S.
can reduce the sense of isolation often felt by
families.

Childcare by experienced carers is provided in a
safe environment catering to the special needs of
T.S. affected children and their siblings.

For further information please contact

Sue Pinkerton

President, ATSS Inc

Ph 9630 3147
