AGSA
THE ASSOCIATION OF GENETIC SUPPOR T OF AUSTRALASIA INC.
NEWSLETTER

CONTENTS

MISSION STATEMENT

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

EDITORIAL

Recently I attended the Smith Magenis syndrome workshop in Wagga Wagga organized by Dr Ingrid Muenstermannwhich from Charles Sturt University. It was a very valuable learning experience which is shared with you in the A_Z genetic condition profile and family story.. Five wonderful women spoke of their experiences. They were just amazing in what they have achieved and what they have to deal with. As one of them said "if my husband was accused of wife bashing, I would get help but when it is my child hitting me, there is no help at all". It was lovely to meet with Brenda Funicane, Executive Director. Genetic Services, Elwyn, a large nonprofit corporation in Media, Pennsylvania, who provide day, residential and consultative services for children and adults with developmental disabilities. (www.elwyngenetics.org) . Brenda has done a lot of work with families on the issues of behaviour and has developed a number of coping strategies which she detailed in her presentation.

In this newsletter I am calling on YOU to wave the flag and write to the ministers or meet with your local minister about respite, schooling and group homes. Things need to change and we can do that by having a united voice. (see articles inside)

Rare Disease Day 28th February 2010 was a united effort with the Smile Foundation, Steve Waugh Foundation and the Australian Paediatric Surveillance Unit. I presented at the APSU and Steve Waugh workshop at the Kids Research Institute on Saturday 27th February. AGSA attended the Smile Foundation fundraiser for Rare Diseases at Boomerang House, Elizabeth Bay on 24th February. Dr Jonny Taitz, Chair, welcomed us highlighting the need to support families with rare diseases. There were many awareness type activities all around Australia. (See further details inside.)

Our **Filling the Void** project has been busy and we are half way through our second Telegroup counselling session for the year. This is a rewarding way to support Mums and families living in rural areas and a good opportunity to share experiences. **Dubbo Carers Seminar** is this month on 20th March at Cascades Motor Inn followed by a sibling workshop the next day. AGSA is working in partnership with MDA, Spastic Centre and Northcott organisations on this seminar.

Mum's Pamper Day 20th April at AGSA from 11am to 1pm. Come along and have a massage, get your nails done and then relax over lunch with a glass of bubbles.

On 27th June we are holding a Dynamic Partnerships Through Change Workshop 10am – 2pm. Lunch will be provided. Come along to explore the challenges due to life changes and techniques on how to deal with them together. Professionals are welcome too.
AGSA is holding its first Adult Sibling Group meeting on 18th April from 9.30 to 1 at AGSA so please let families know.

I look forward to hearing from you.
Until then
Best wishes

DIANNE PETRIE

Call to Action: KINGSDENE SCHOOL

AGSA is asking for your help in drawing the attention of your school's community to the article regarding Kingsdene Special School in the March issue of Marie Claire magazine
and especially to the on-line petition to save the school which will be presented to Mr Bill Shorten. The story and petition can be found at

http://au.lifestyle.yahoo.com/marie-claire/features/reports/article/-/6741343/the-em-battle-em-for-care-thats-pulling-families-apart/

PRESS RELEASE
Call to Action:
NO GROUPS HOMES NO ONGOING CARE

For Immediate Release 10.20am, March 1st, 2010
Lynch Lies and Plays Catch 22 With Ageing Carers of Intellectually Disabled

The parents of a group of intellectually disabled adults in Sydney's Ryde area are horrified that Paul Lynch, the NSW Minister for Ageing, Disability and Home Care (ADHC), has denied he ever gave in-principal approval to their innovative plans for supported accommodation for their 20 sons and daughters.

According to an article in today's *Sydney Morning Herald*, http://bit.ly/bUkaRC "Mr. Lynch told the *Herald* the plan would be considered once land was available. '… I did not give in principle support to the RASAID proposal, as the land has not been secured and there would be numerous other steps which would have to be undertaken.'"

He also denied signing his name in the air after he'd given Jenny Rollo, now President of RASAID, verbal support of their plans. Ms Rollo says there were several witnesses to the aerial signature, at least one of who is prepared to swear in a statutory declaration that her version of events is correct. The others are members of the NSW Labor Party or employed by Minister Lynch. Commenting on this "signature" to Ms Rollo and another RASAID mother on the day it occurred, Lynch's aide said, "If Minister Lynch wants this to happen, it WILL happen."

But worse than being called a liar, Ms Rollo is outraged that the minister says the group's model would be considered once land is secured. "He's set up a Catch-22 situation," she says. Land owned by the NSW Department of Health in the Ryde district was identified as being available to RASAID in early 2009. At a meeting on June 16th 2009 between senior bureaucrats from the NSW Department of Health, the NSW ADHC, RASAID and the NGO chosen to build the project, an acre of land was to be offered for this project.

"The senior bureaucrat from ADHC told us at that meeting that even if our cluster housing was built, there was no guarantee that all, or in fact that any of our children would go into it because of the department's Vacancy Management Policy. The plans of the land were then removed from the table, literally and figuratively," says Ms Rollo.

ADHC's Vacancy Management Policy stipulates that the department decides who goes into what available beds in NSW, rendering RASAID's plans for an intentional community for its adult children with intellectual disabilities impossible.

Undeterred, Ms Rollo and another RASAID mother wrote to Minister Lynch, asking for him to meet with them and to override ADHC's policy to allow their proposal to go ahead. The minister refused a meeting and said, via his staff, "We have no further developments to advise RASAID on at the moment."

On November 2nd last year, four months after the original meeting when the land was to be offered, Ms Rollo received confirmation via the ADHC that the Department of Health land was still available for their project.

"It beggars belief that he now says he'll consider our proposal once land is secured. Land can't be secured unless he makes an exception to his department's policy. It is that alone that is preventing the land from being offered," says Ms Rollo.

RASAID is calling on Premier Kristina Keneally to step in and remove the bureaucratic barriers that are preventing RASAID's development from going ahead.

RASAID's supported accommodation model falls within ADHC guidelines for a cluster development to house intellectually disabled people. The model proposes that RASAID's sons and

daughters live together within the Ryde area, with their friends, near their families, day programs and work placements. The current system for housing intellectually disabled adults who can no longer live with their ageing or dead parents is ad hoc. The state meets only 7% of supported accommodation needs of adults with intellectual disability.

RASAID members are in their 50s, 60s, 70s and 80s. The oldest parent is a single mother, aged 87, still caring for her 51 year old son.

~~~~End of Release~~~~

For all media enquiries and to arrange interviews with Jenny Rollo and other RASAID parents, please contact Teresa Russell on 02-9807.9693 or 0400 00 23 49 trussell@aapt.net.au

A RASAID family, Dick, Marilyn and Robbie Jones, was recently featured on *Four Corners'*program, *Breaking Point.* This is a sample of the families the minister is hurting with hisCatch-22 policy web. http://www.abc.net.au/4corners/default.htm

Ryde Area Supported Accommodation for Intellectually Disabled Inc. ABN 87 321 781 096 DGR endorsed PO Box 2068, Boronia Park, NSW 2111 ph: 0408 970.806 rasaidgroup@gmail.com www.rasaid.org.au

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Article from Sydney Morning Herald today 1st March 2010

Housing scheme for disabled adults in doubt as minister backs down

ERIK JENSEN
March 1, 2010

EIGHTEEN months ago two women sat in a Gladesville kitchen and pleaded with Paul Lynch to approve a housing scheme for their disabled adult children. What happened next is in dispute.

They say the Minister for Disability Services reached forward with his finger and drew his signature in the air. After six years of lobbying, his gesture was the closest their cluster housing scheme had come to approval. The minister denies it ever took place.

"'We left that meeting feeling that it would actually happen," the group's president, Jenny Rollo, says. "It was the same feeling we had with Kristina Keneally because she was so supportive." But the RASAID scheme, a residential care facility for the disabled children of a group of older parents from north-west Sydney, is no closer to approval under Mr Lynch than it was when the Premier held the portfolio.

"I do not," the minister says, "draw my signature in the air." Talking to Ms Rollo, ministers walk in and out of conversation. Almost 60 meetings with politicians and bureaucrats in the past six years. The project's plans sit on her table, but need government funding for construction and ongoing care. "We're made to feel that the best solution is for our kids to die before us," she says.

"One of the hardest things about being the parent of an adult with a disability is not being able to plan for the future. It doesn't matter if you are rich or poor - there's no way to plan. The state government expect our other children to take over."
Since the group of 19 families formed, two parents were forced to relinquish their 32-year-old son because his needs had become too great. "We're still going through the whole process," his mother says, "of coping with what we've done."
While there is some hostility to the scheme, with British research suggesting accommodation of this type can be restrictive, each family believes it is important to keep their adult children together. Many have known each other for 30 years but current systems of care would give no guarantee on where they end up once they leave their parents' homes.

Mr Lynch told the *Herald* the plan would be considered once land was available. "... I did not give in-principle support to the RASAID proposal, as the land has not been secured and there would be numerous other steps which would have to be undertaken."
END STORY

Every family, their parents, siblings, cousins, next door neighbours and friends – everybody you know: ask them to write.

Q - How can I send a Letter to the Editor? (Print edition)

A - *The Sydney Morning Herald* receives over 400 emails each day as well as faxes and snail mail for Letters to the Editor. This makes it impossible to reply to all correspondence. The Letters Editor can only publish 35 - 40 letters on the page. All letters and email (no attachments) to the Herald must carry the sender's home address and day and evening telephone numbers for verification. Ideally, letters will be a maximum of 200 words. By submitting your letter for publication, you agree that we may edit the letter for legal or space reasons and may, after publication in the newspaper, republish it on the internet or other media.
Email: letters@smh.com.au
Mail: GPO Box 3771, Sydney NSW 2001
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Phone	(02) 9228 5239 (02) 9228 5239
Fax	(02) 9228 3934
Email	thepremier@www.nsw.gov.au

Local people can show their support by writing to our local MP, Vic Dominello, who has publicly stated that he will build the RASAID houses "when the libs are voted in at the next election":

Address	Mr Victor Dominello, MP Level 1 89 Blaxland Road RYDE NSW 2112
Phone	(02) 9808 3288 (02) 9808 3288
Fax	(02) 9877 6222
Email	ryde@parliament.nsw.gov.au

Or to the Shadow Minister for Disability Services, Andrew Constance, who also supports RASAID.

Address	Mr Andrew Constance, MP 122 Carp Street BEGA NSW 2550
Phone	(02) 6492 2056 (02) 6492 2056
Fax	(02) 6492 3578
Email	bega@parliament.nsw.gov.au

We would like your opinion

Taverner Research is conducting a Client Satisfaction Study on behalf of the NSW Department of Justice and Attorney General of people with disabilities who have dealt with any of these NSW services in the past twelve months:

- The Anti Discrimination Board
- The Administrative Decisions Tribunal
- Law Access
- A Local Court
- NSW Trustee and Guardian (clients of the former Office of the Protective Commissioner)
- Registry of Births Deaths & Marriages
- Victims Services

The outcomes of the study will help to improve these services for people with disabilities. As a thank you for taking part in the study participants will receive a $75 gift voucher.

If you would like to take part or would like to know more about the study please call Richard at Taverner Research on 1800 212 290 or email him at dsurvey@taverner.com.au.

Or go to this web address for more information: http://www.taverner.com.au/surveys/w3571.htm

BIONEWS

Reprinted with permission from BioNews (www.bionews.org.uk)"a free web-and-email-based news service on human genetics and assisted reproduction, published by the UK charity Progress Educational Trust.

Patients put off genetic testing over insurance fears, study shows

14 September 2009

By Rosie Beauchamp

Appeared in BioNews 525

A study published in the Medical Journal of Australia indicates that up to 50 per cent of Australians could be refusing genetic tests due to the fear that the test outcomes will result in increased health insurance premiums or refusal to insure them altogether. In Australia, genetic information does not inform health insurance. However, it may affect insurance policies that cover trauma, disability, sickness and accidents. Applicants for such health insurance policies have a statutory duty to disclose known relevant information and so long as insurance companies comply with anti-discrimination law, they are within their rights to refuse insurance, or charge a higher premium based on the information provided.

The study, carried out at Melbourne University, identified 106 people from 25 different families thought to be at increased risk of bowel cancer. When the participants were initially offered the screening tests 80 per cent were in favour. However, the number willing to find out the results of their tests decreased by 50 per cent when participants were informed that insurance policies may be affected by the results.

In Australia bowel cancer is the second most common cancer in both men and women, with one in 3000 Australians having a genetic mutation putting them in a high risk category. Screening for bowel cancer is an effective way of diagnosing and removing polyps from those with the genetic mutation, and thus preventing the disease.

Senior author of the paper Mark Jenkins said: 'Insurance-related apprehension about genetic testing could have troubling public health consequences. Screening people at high genetic risk of bowel cancer is a highly cost effective way to reduce deaths due to bowel cancer'.

In response to this research, the authors of the study have called on the Federal Government and Australian insurance companies to reconsider the current policy. The paper concluded: 'If the industry's position on genetic information deters individuals from obtaining test results, the clinical and public health consequences could be damaging'.

Canada considers genetic discrimination law

01 March 2010

By MacKenna Roberts

Appeared in BioNews 547

Canadians need better protection from genetic discrimination by insurers and employers, according to Winnipeg North MP Judy Wasylycia-Leis.

Mrs Wasylycia-Leis announced she will introduce a proposal into the Canadian House of Commons this spring amending the Human Rights Act to include 'genetic characteristics' as a prohibited ground for discrimination. 'This bill will stop Canadians' personal genetic information from being used against them', she said.

Canada needs to follow other nations' legislative initiatives, Mrs Wasylycia says. She reportedly believes abuse of genetic information will only increase without regulatory safeguards as more and cheaper genetic tests become widely available to detect a person's susceptibility to a growing range of genetic conditions.

'Unless genetic test results are protected, there's a real danger that Canadians will just refuse to be tested, putting their health at risk', she said.

The Canadian Coalition for Genetic Fairness (CCGF), a coalition of 15 disease-related associations dedicated to preventing genetic discrimination, helped Mrs Wasylycia-Leis draft the bill. The CCGF wants regulatory reform for the insurance industry in light of genetic advances.

Mr Don Lamont, CEO of the Huntington Society of Canada and chair of the CCGF, says predictive testing is 'a good thing', but warned it also carries 'a growing fear that the information can lead to stigma and discrimination'.

The bill would not explicitly prohibit insurers or businesses from demanding genetic tests, but will allow the courts to decide what practices are discriminatory. Mr Lamont believes it should also invite judicial examination of privacy legislation and genetic information.

The CCGF estimate that the number of genetic tests available over the last decade has increased from 100 to over 1,500.

Many nations are currently examining and regulating on genetic discrimination. In May 2008, the US passed

legislation prohibiting employment and health insurance (not life insurance) genetic discrimination.

In the UK, there is a voluntary moratorium between the government and insurance industry on the use of genetic test results until 2014. Exceptions include testing for Huntington's disease in patients requesting life insurance above £500,000 or health cover above £300,000.
In comparison, France and Spain strictly outlaw all use of genetic test results by all insurers.

Welcome to Rare Disease Day 2010 in Australia 28th February

Families rally around Australia to raise awareness of rare diseases.
It is estimated that the ~8000 known rare diseases affect ~1.2 million Australians and have wide-ranging impacts on families, health professionals, health services and the community.
The following organisations will hold activities to raise awareness of rare diseases in Australia.

The Association of Genetic Support of Australasia Inc. (AGSA), formed in l988, is an umbrella group providing support and information for individuals and families affected by a genetic condition. AGSA is working together with our members to raise awareness of Rare Disease Day and is promoting events held around Australia. www.agsa-geneticsupport.org.au

Australian Paediatric Surveillance Unit has been conducting research into rare childhood disorders since 1993 www.apsu.org.au and in February 2009 convened a National Working Group to develop a co-ordinated approach to address the impacts of rare diseases in Australia.
The SMILE Foundation is an Australian charity that helps families whose children suffer from rare diseases. SMILE is holding a cocktail event in order to raise funds for its Family Relief Program and awareness in the community.

SMILE is assisting parent groups of rare diseases around the country to hold functions in honour of IRDD.

SMILE will also be creating media opportunities via the families we have helped in the last 2 years.

Steve Waugh Foundation:
"We have been supporting & helping Australian children and families since 2004. I know I really can make a difference and bring hope to children and families affect by rare diseases. To me being Australian is about looking after your mates, taking care of the less fortunate, supporting the underdog and enhancing the spirit that makes all Australian's unique." Always 100% ™
(Steve Waugh, AO)
The Steve Waugh Foundation looks forward to working with all charities and organisations that support children and families affected by rare diseases in Australia and we look forward to being a member of the Australian team in this arena.
www.stevewaughfoundation.com.au
For any media enquiries please contact:
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NDIS
the Productivity Commission inquiry into a National Long Term Care and Support Scheme for people with a disability and their families.

Last week the Productivity Commission received their terms of reference for the inquiry – you can view the terms at www.pc.gov.au/projects/inquiry/disability-support/terms-of-reference.

The terms of reference are deliberately broad and wide ranging to allow a thorough exploration of the many complex issues which would be thrown up by the introduction and implementation of a National Care and Support Scheme.

The Productivity Commission will hold public consultations as part of its inquiry. It is absolutely vital that the Commission hear from people with a disability, their families and carers, as well as the organisations that support them. If the system is truly to be reformed it must take account of the experiences and needs of people with a disability and their families. It is only with these experiences as a base that real transformative change will be achieved. The commission must hear the voices of those with real lived experience of disability.

This is your chance to have your say about how you would like the system to change - do not miss this important opportunity to shape the future of people with a disability, their families and carers in this country.

Make sure you log on to the Productivity Commission website and register your interest. That way you will know when the consultations begin and how you can participate.

News from NORD's Newsletter February 2010

Millions Around World to Observe Rare Disease Day on Sunday

Sunday, Feb. 28, will be the third annual World Rare Disease Day. Patient organizations and others around the world will observe the day with gatherings, events, and online postings to raise awareness of rare diseases as an important public health issue. This year, there is also a focus on the need for more research to develop treatments for the many rare diseases that currently do not have them. The special relationship that exists between patients and rare disease researchers also is being emphasized this year. To read about plans for Rare Disease Day in the U.S., go to www.rarediseaseday.us . For a global overview, go to www.rarediseaseday.org.

Database of Rare Disease Experts. As part of the Rare Disease Day observance, NORD has launched a project to create a database of physicians with expertise on various rare diseases. To nominate a physician (either a clinician or researcher or both), provide basic information on this online form. NORD will contact the physician to determine whether he/she is interested in being included in the database and to gather additional information.

NORD and EURORDIS Launch Advocacy Blog

NORD and the European Rare Disease Organisation, EURORDIS, recently launched an international advocacy blog at www.rarediseaseblogs.net . Invited bloggers include experts from the U.S. and Europe on topics related to rare disease research and the development of orphan products. Recent posts include one from NORD President Peter L. Saltonstall on the recent expansion of the Social Security Compassionate Allowances program and one from EURORDIS CEO Yann LeCam entitled "Rare Diseases at the Forefront to Address the Healthcare Challenges of the Future."

Everyone with an interest in rare diseases and orphan products is invited to visit the blog often and post comments.

OVERSEAS NEWS

GENETIC ALLIANCE USA NEWS

Rare Disease made the final round for the 2010 Change.org Top Ideas Contest. This is really wonderful. We beat out hundreds of ideas to make the finals and now must reach the top 10 out of 60 final ideas.

Voting starts Monday, March 1 at 1 p.m. EST through Friday, March 12 at 5 p.m. EST. Please vote – then we can easily rise to the top of the pack!!

This is truly an amazing chance to make rare disease a health priority in the USA (and world).
There are more than 25 million people in the USA alone with rare disease with millions of family members touched by rare disease as well. Pseudoxanthoma elasticum (PXE) represents only about 10,000 individuals, but working together will all of the other rare conditions we get so much more done.

Please vote at Change.org – http://www.change.org/ideas/view/25_million_it_is_time_to_care_about_rare_disease

Press Release

For Immediate Release

International Alliance of Patients' Organizations
A global voice for patients

Patients call for greater support for meaningful involvement in healthcare decision making as a means to ensure equitable distribution of limited health resources

- *A Call from IAPO's 4th Global Patients Congress on Strengthening Healthcare Systems Globally* -

London, United Kingdom, 1 March 2010 – Patient advocates have called for more meaningful engagement in healthcare design and delivery at the **International**

Alliance of Patients' Organizations (IAPO) 4th Global Patients Congress in Istanbul, Turkey. The Congress was held with the support of the Access to Health Association of Turkey.

Patients' organizations are knowledgeable and motivated to work with information, facts and figures to promote policy change, providing they are involved at decision-making level and treated as equal partners. Patients add value to discussions and decisions by enabling analyses that take into account the reality of what is happening within health systems today. The impact of the global economic situation on healthcare budgets and changing demographics, including a dramatic increase in the number of people with chronic conditions, requires health systems to change and adapt rapidly. Patients must therefore be at the centre of healthcare globally and at the forefront of decision-making.

"We have identified models of patient and public engagement that are successful and can serve as catalysts for further change within healthcare systems. One such model is the European Medicines Agency, where patients play an active role in a number of committees. We strongly advocate that patient engagement should happen in all health systems to the benefit of systems, patients, and thus the public at large." says Hussain Jafri, Chair of IAPO. "Real concerns can be aired and appropriate measures taken that empower patients, health professionals and authorities alike."

The Congress brought together over 100 delegates from around the world, representing patients and other stakeholders in health such as the Council of Europe, the International Council of Nurses (ICN) and the World Health Organization (WHO) to address the subject of **Strengthening Healthcare Systems Globally: The Value of Patient Engagement**. Patient engagement has been a principal focus of IAPO's work since its foundation. IAPO advocates for patients and patients' organizations to share the responsibility of healthcare policy-making through meaningful and supported engagement at all levels and at all points of decision making. IAPO is a unique global alliance representing patients of all nationalities across all disease areas and promoting patient-centred healthcare around the world.

More information on the Congress can be found at www.patientsorganizations.org/congress
IAPO's Policy Statement and Guidelines on Patient Involvement can be found here
For further information, please contact:
Rebecca Buckley, Admin and Information Officer, IAPO, T: +44 20 7721 7508; Email: info@patientsorganizations.org

CONFERENCES

CALL FOR PAPERS 2-4 August – Association of Children's Welfare Agencies Conference 2010: Sydney Convention and Exhibition Centre. Call for papers are now open and will close on 25 March 2010. Guidelines for submissions at http://www.acwa2010.com/abstract.asp

Third Annual National CJD Conference

Rydges on Swanton – Carlton Victoria
Saturday 15th May 2010

An afternoon of Information on CJD
Sunday 16th March 2010
Kirribilli Club – North Sydney
11 Harbourview Crescent, Lavender Bay
1pm Registration for Health Care Professionals
Cost: Free of charge

Understanding CJD
Thursday 13th May 2010
Venue: Mantra on Murray
305 Murray Street, Perth WA
For more information
Suzanne Solvyns
Director
National Coordinator
CJD Support Group Network
TelFax +61 2 98998905 Toll Free 1800 052466
email: s.solvyns@cjdsupport.org.au
website: www.cjdsupport.org.au

A – Z GENETIC CONDITION PROFILE

SMITH-MAGENIS SYNDROME

- Includes: Chromosome 17, Interstitial Deletion 17p, Smith-Magenis Chromosome Region, SMS

Smith-Magenis syndrome (SMS) is a rare chromosomal deletion condition in which the symptoms include distinct features of the head and facial (*craniofacial*) area, delays in the acquisition of skills requiring the coordination of mental and muscular activities (*psychomotor delay*), cognitive delay, and/or behavioural abnormalities (such as

self-destructive behaviour and hyperactivity). In addition, most individuals with SMS experience speech delays that may occur in association with hearing impairment and have an abnormally hoarse, deep voice. Many also experience sleep disturbances including difficulties falling asleep and staying asleep.

SMS is caused by a deletion (missing part) of the short arm (p) of chromosome 17(17p11.2). The chromosomal deletion occurs due to a spontaneous (*de novo*) genetic change that occurs for unknown reasons (*sporadic*).

WHAT IS SMITH-MAGENIS SYNDROME?

Babies with Smith-Magenis syndrome (SMS) typically have unusual facial features that may include a broad square-shaped face, an abnormally short, broad head (*brachycephaly*); an abnormally broad, flat midface; a broad nasal bridge; an unusually prominent jaw (*prognathism*); eyebrows growing across the base of the nose (*synophrys*); a short full tipped nose and fleshy upper lip with a tented appearance. The head appears short and flat. Most children with SMS have a raspy or hoarse voice, and approximately three quarters have speech delays and a loss of hearing. They may also have short, wide fingers and toes.

Growth delay and intellectual impairment, as well as hyperactivity usually occur in children with SMS. Measured IQ can range from 20 to 78 with most individuals falling in the moderate range of intellectual impairment, between 40 and 54. Self-destructive behaviours occur in two thirds of individuals who are affected with SMS, and may include head-banging, wrist-biting, the insertion of foreign objects into the nose and ears (*polyembolokoilamania*), and pulling out the nails of the fingers and/or toes (*onychotillomania*). Extreme nearsightedness (*myopia*) and crossed eyes (*strabismus*) frequently occur in children with the condition. Other visual problems may be the result of retinal detachment (separation of the retina to its attachment at the back of the eyeball). Around three quarters of children have some form of eye abnormality.

Children with SMS may experience significant sleep difficulties (75%) including falling asleep and/or remaining asleep. Some children may have a decreased sensitivity to pain, burning sensations, loss of feeling in the legs (*peripheral neuropathy*), loss of muscle mass in the legs (*amyotrophy*), and absent or decreased reflexes.

Other less common features of SMS include cleft palate, *cardiac* (heart) defects, curvature of the spine (*scoliosis*), brain abnormalities, *renal* (kidney) problems, forearm abnormalities and immunological problems.

WHAT CAUSES SMITH-MAGENIS SYNDROME?

Genetic causes of Smith-Magenis syndrome

SMS usually occurs due to an absence of genetic material (*interstitial deletion*) on the short (p) arm of chromosome 17, specifically at 17p11.2. Several genes are contained in this part of the chromosome. Figure 1 below is a diagram of chromosome 17 with the relevant area arrowed.

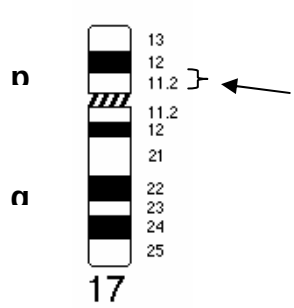


Figure 1 – Chromosome 17

Chromosomes are found in the nucleus of all body cells. They carry the genetic characteristics of each individual. A deletion is the absence of part, or all of a particular area of a chromosome. Symptoms vary according to the amount of missing genetic material. Children with a smaller deletion may have less severe symptoms than those with a larger deletion of that area on chromosome 17. In individuals with SMS, the chromosomal deletion occurs due to a spontaneous (*de novo*) genetic change (*mutation*) that occurs for unknown reasons, and is therefore said to be *sporadic*.

It is sometimes not possible to identify a deletion at 17p11.2 in individuals with SMS. In these people, it is possible that they may carry a faulty copy of the *retinoic acid-induced gene (RAI1)*, located in the same part of chromosome 17. This faulty gene is inherited in an autosomal dominant manner. This may, however, be a new genetic change (*de novo*) in the individual with SMS, and not inherited from a parent.*Autosomal dominant inheritance*

Autosomal dominant inheritance refers to the inheritance of a 'dominant' gene change (*mutation*) located on an autosome (one of the numbered chromosomes). There are two copies of every autosomal gene. Both copies of the gene send a message to the body, but a dominant faulty gene copy will override the message sent from the working gene copy.

If a parent has an autosomal dominant condition, there is a 50% chance that they will pass on the dominant faulty gene (and therefore the genetic condition) to each of their children. This chance is the same for each pregnancy and is the same for males or females.

For more information about genes, chromosomes and the autosomal dominant form of genetic inheritance, please refer to Genetics Fact Sheets 1, 2, 4, 5, 6 and 9.

WHO IS AFFECTED BY SMITH-MAGENIS SYNDROME?

SMS is a rare chromosome condition that affects males and females in equal numbers. The estimated prevalence is between 1/15,000 and 1/25,000 births, though this may not reflect figures for Australian populations. SMS has been identified worldwide in people of many ethnic groups.

HOW IS SMITH-MAGENIS SYNDROME DIAGNOSED AND TREATED?

Diagnosis

A specialist such as a clinical geneticist may make a diagnosis of SMS in a child who has the clinical features of the condition. These features can be subtle in infancy and early childhood, frequently delaying diagnosis until school age, when the characteristic facial appearance and behavioural phenotype may be more obvious.

SMS is diagnosed by the detection of a deletion of the short arm of chromosome 17 at band p11.2. This deletion can be identified following a routine chromosome analysis but is sometimes overlooked. Specialised chromosome testing using a technique called *fluorescent in situ hybridization* (*FISH*) may be necessary to show extremely small deletions that cannot be seen under the microscope.

Specialised molecular DNA testing via a blood test may be helpful for identifying an *RAI1* gene mutation in a small proportion of people with SMS, particularly if it is not possible to find a deletion on chromosome tests.

Treatment

Recommended therapies include speech/language, occupational, physical and behavioural. Psychotropic medications may be useful in increasing attention and decreasing hyperactivity. Melatonin therapy may be helpful for the sleep disorder. Early intervention, special education programs and vocational training are recommended to maximise the potential of those with SMS. Respite care and family support may be important to help assure the optimal environment for the affected individual.

Genetic counselling may be of benefit to individuals with SMS and their families.

Genetic Testing Options

Genetic testing may be available for this condition. Genetic testing may be carried out on individuals, on an unborn baby (*prenatal testing*) or on an embryo (*preimplantation genetic diagnosis*). For the most appropriate and accurate information, contact a genetic counselling service to find out whether genetic testing is available for this condition and discuss your specific options and questions.

Additional information and Support may be available from the following sources:

Smith Magenis Syndrome (SMS) Australia
3 Lilli Pilli Point Road
LILLI PILLI NSW 2229
Ph: (02) 9524 9902
Email: kate_tye@hotmail.com
Web: www.gsnv.org.au/SMS/

The Smith-Magenis Syndrome Foundation
24 Brook Road, Dersingham
King's Lynn, Norfolk PE31 6LG, UK

Ph: +44-(0)132-8730-782
E-mail: info@smith-magenis.co.uk
Web: www.smith-magenis.co.uk

FAMILY STORY

SMS and the impact on Siblings

A lot is documented on the maladaptive behaviours of SMS. Opposition, defiance, aggression, obsession, destruction, outbursts, meltdowns. These are all great adjectives to describe violence. That is the way children see it anyway. Our siblings don't have the fancy descriptions; all they have is an innate sense of fear, an understanding of the dangers involved with being close to

their brother or sister and the knowledge that their life is different from others, unsafe, dangerous and isolated. They find safe places in the house to hide, places where they are sure they will never be found, places to hide precious things they don't want broken and ways to manage their heightened stress levels.

Sometimes they don't manage them at all and get labeled by their peers and others as different.

Of course these children are different! They live in a strange world, none of their other friends could understand. We as parents are stressed to the point of dysfunction and often are unable to give as much as our very needy siblings require. I know I tried my hardest but then my body broke and there was nothing I could do about that, except rest and cry and take steps to repair.

Our Story

By the time Lachlan was 10, he had managed to get a tight grip on our family life. At first we had tried to get around his bad behaviours as each one cropped up, we had set up a bedroom for his nocturnal goings on, we had close circuit TV in the kitchen with cameras throughout the house so I could monitor him and still perform basic household chores, and we had a strict routine, which had to be followed or there would be trouble. Lachlan hated leaving the house; his main focus was keeping the TV on Nick Jr and having his computer on at the same time so he could go back and forth at his leisure. For a while when Tim and Emma were little, this seemed like a sensible pattern.

What it did though was tighten a noose around our neck that we could not loosen. As he got older, he tightened it up harder so that we had to be in the lounge room with him, he had to control us too. If one of us left, he would make us pay. If I went to the bathroom, he would quickly get up and go straight to Emma's room and break her things. Of course this meant Emma would go in behind him to protect her stuff and would invariably get hurt. She has had a broken arm and a split lip from such times, but this would then get me back into the room. Even if I was angry, it still meant he had control of me.

So by the end, I could not leave the lounge room without Tim and Emma. I had to keep them in my sights to know that they were safe. I can assure you this is no way to live. But even then we were given little support. Out of home respite down to one weekend in 12 or so and one night per fortnight. It was not enough, it never is. We had psychological assessments that talk about his behaviours in the pervasive range. Fully documenting what was going on. All those adjectives being used instead of the word Violence. We were constantly living in fear and no one was prepared to listen and take it for what it was. We were terrorised in our own home. ADHC had removed our case manager, because all our problems could not be fixed.

For Lachlan the fact that I was taking Tim and Emma with me through the house meant that he had lost control of me. So he decided to ramp it up and started mutilating himself. This I could have no control of and it gave him all the control. Our house was covered in blood. If CSI came with their luminescence thingy our house would light up like a Christmas tree! He started by pulling his teeth out, ready or not, then by putting things up his nose, by picking at his skin, by biting himself and then the final act by pulling his Gastrostomy out. Not only was my house covered in blood, wee and faeces but now there was stomach acid too.

I took him to the hospital because I couldn't get him to keep the gastrostomy in and I was shocked by the lack of understanding. One Doctor suggested the use of cable ties to restrain him. In this environment, I became very tired, I realised that I hadn't slept in days and I hadn't really eaten. I was just exhausted and an uncaring nurse pushed me too hard and I had a little psychotic episode of my own. Then they were looking for a bed for me (in the psych ward) but of course that would mean they would also have to admit Lachlan and they had decided he would be too hard to look after, he would take up too many resources, so I would just have to take him home and the sooner the better. I would not move though, I knew that I was not fit to care for Lachlan in that state. Docs told me to take him home, they had nothing to offer us. In the end I found the emergency bed at respite. That is where he currently resides some 18 months later. There are no options for us. There are no group homes for children. I managed to get him a place at Kingsdene Special School, a wonderful place that boards about 20 kids in similar situations to ours. So he lives there Monday to Friday and still is at respite on the weekends.

Since Lachlan has left home, I have had time to see the impact he has had on all of us, and this is the story of Tim and Emma's journey.

Tim's Journey

When Tim was born, Lachlan was 3 and a half. Lachlan tolerated a new baby well enough but by the time Tim was 2 and getting around he was less tolerant. He would hit or push Tim out of the way if he came too close and a few times managed to push him down the stairs.

I can remember as a 3 year old Tim asking me "why does Lachlan hit me?" Hard to explain but it was not long after this that we finally had a diagnosis of SMS and that gave me a good answer. "It's not Lachlan that hits you it's his

SMS". So some days I could say Lachlan's SMS is really bad today, please stay out of his way. And Tim really knew he had to be careful.

Then when Emma was born he would make sure she stayed out of harms way too. He would take her to his bedroom and find things to amuse her. At this point someone pointed out to me that he had become a young carer.

We also had artwork from preschool that showed our life, me bashed and Nigel rushing to my rescue. It was a clear and accurate depiction of our lives. One night after a particularly bad evening Tim sat next to me on the lounge and patted my knee, "sometimes it's really hard living with SMS, mum". A young carer, caring for his mother too.

When Tim was 5, he started asking questions about the future, I told him that one day I would get too tired to look after Lachlan and he would move out of home. Tim told me not to worry, that by that time, he would be old enough and he would take care of Lachlan. I was horrified to think that he had already decided that was his future. I had never said anything that would make him think Lachlan would be his responsibility. He is a deep thinking boy and he had already seen the need for Lachlan to be looked after for his lifetime.

At this time Tim had started school and it became apparent that he lacked the social skills to fit into the playground. He actively avoided rough and tumble playground games and cried easily and became the target for bullies. He was set upon by 3 kindergarten children at one point and was kicked and bashed to the ground and needed to go to hospital. In the classroom, he spent most of the day staring out of the window or hiding under the desk, crying. At this point I took him to see a psychologist who became aware of the violence in our home.

If she had discovered that it was my husband who was violent, then the police would have been called, we would have had resources thrown at us and my husband would have been in jail. Instead, because it was a child who was the perpetrator, Tim was given counselling on how to be brave, to take control of his "worry monsters". We were given instructions on how to write a social story that I would read to Tim every night.

This social story included ways to stay safe, where he could be safe and if he was too frightened when I was being hit and the screaming became too loud, how to close his eyes and imagine being somewhere that he had enjoyed in the past.

Back then it seemed like such a sensible thing to do. Now I am appalled to think that health professionals thought this was appropriate, and could not see our lives for what they were.

We were living with domestic violence.

The powers that be talk about teaching the family resilience. What exactly does that mean? How do you teach resilience to a 5 year old who gets bashed every day? No one expects a 5 year old to just get over it and learn to be brave if an adult physically abuses them. Why is it that we had to be resilient to the same level of violence just because it was perpetrated by a child? A situation that has left both Tim and Emma long term scars that will need a lot of help to heal.

Since Lachlan has left home Tim has had extensive counselling. By listening to the counsellors, I have been given some understanding of the way he thinks, and ideas to help him into the future. To help him heal.

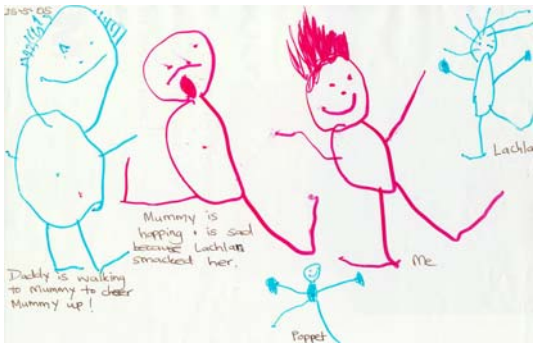
I have found out that when he was in his room hiding from the violence, he was worried for my safety and spent the whole time thinking that he wasn't brave enough, strong enough or good enough to help me.
All this time I thought he was safe, but he was having his self esteem destroyed.

I have found out that all the time Tim and Emma were playing in the backyard it was because they knew Lachlan would never come outside and they would be safe.

They had developed plans to maximise their own safety.

It is only with time that he is beginning to be able to verbalise some of the trauma that he has suffered by the hands of his brother.

Tim still has a difficult time in the playground at school. It is my belief that both Tim and Emma are well behind their peers on an emotional level.



Tim was asked to draw his family at preschool. He was 4 years old, Lachlan had just turned 8. It was around this time we had carers refuse to come to our house and look after Lachlan.

Emma's journey

Emma has had Tim to help her through, but she has still suffered a great deal. She has had broken bones and broken skin. She has been grabbed, pushed, kicked, pinched and had her hair pulled. The last time Lachlan hit her, he punched her in the face and split her lip, blood everywhere, she never really forgave him for that one. After that I wouldn't leave her alone with him.

When Lachlan left home, I put Emma in counselling too. She was much more open to counselling than Tim and her progress was so interesting to watch.

On the first day, she found the wet sand tray, found a little figurine that she labeled "the scary monster" and proceeded to bury that under every grain of sand available. A huge volcano like mound. Every week she would came in find the scary monster and bury it. Over time the scary monster became the naughty brother and we were represented too, little figurines all placed around the base of the volcano.

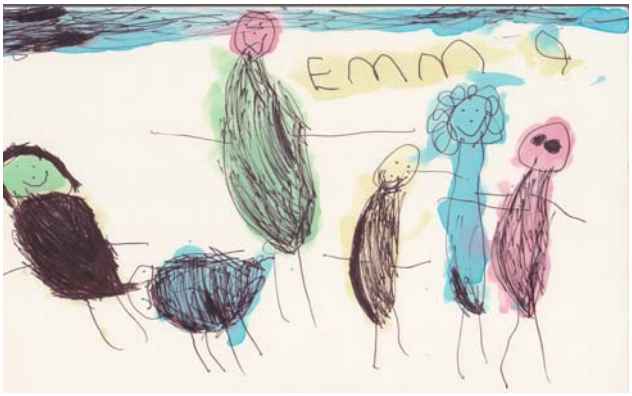
Months passed and the sand volcano became less severe, then became a castle, still the naughty brother was buried. There was a moat and crocodiles in it.

After about 10 months of weekly visits, the wet sand tray was a beach scene with all of us enjoying ourselves and Lachlan with his own name.

The therapist said it was interesting that at times when she got too sad she would break off and draw cards with love hearts on them for Lachlan. She was so conflicted with feelings of love, hate and fear.

Recently she has shown signs of needing to return to therapy. She is prone to anger and when she is confronted with something she is not happy with or worried by, hides under beds, in cupboards, behind doors, under tables. You name it, she has hidden in it. At school she is having problems socially, finding it difficult to manage conflicts and negotiations with her peers. She is emotionally very fragile.

Once again I need help, to help her through the ongoing legacy of living with SMS.



This was Emma's preschool family photo. Lachlan looks so spooky in this picture. It was also pointed out to me that he is red, often used by children to signify aggression. It is interesting that Dad's head is red also. He often had to raise his voice to get Lachlan to behave. I'm also attached to Lachlan, and Emma is removed from us.

This Family picture was drawn at the end of last year, 18 months after Lachlan had left home. Emma has moved

back in the family circle. We are surrounded by happy faces. Lachlan is missing, but that is ok for the moment if it makes her feel better.

The Future

Since being out of home, Lachlan's anxieties have diminished greatly, he is far happier. He has a very structured life, whether at respite or at the boarding school he attends 5 days a week. Structure, familiarity with routine, and a calm caring environment bring out the best in him. He can be such a joy to be around! I would never have said that when I was living with him.

When the children get together, it is at the same location, at a local McDonalds, highly organised so Lachlan is comfortable and happy. Then Tim and Emma can have some time with their brother that is positive, free of violence and without fear.

This is so that in the future they can have a good relationship with him. For Lachlan having a good relationship with his siblings will be incredibly important, after we are gone they will be making decisions for him.

These organised dinner dates have meant a lot for our family and we finally have some pictures of the children together, something that never happened when Lachlan was at home. This is a sign for me that we are on the right track and we can have some hope for the future.

Tim and Emma still require counselling and that is something that will continue some time in the future. The years of living with a frightening unpredictable person, isolated from normal life have taken their toll.

In legislation nothing allows for out of home care for children. Legislation states that all children should reside in a family setting.

What about children that are incapable of living harmoniously in a family setting?

What about children who need so much structure that only a group home setting can give them the scaffolds they require?

The government and the community need to understand that some disabilities mean that all the family is at risk. The idea of "increasing resilience in the family" by ADHC is misguided at best.

The behaviours exhibited by Smith Magenis Syndrome compromise the capacity for families to care effectively and safely for all members and the damage being done to siblings, parents and families is enormous.

Now that Lachlan no longer lives with us, Tim and Emma have the opportunity to form a relationship with him based on Love and happiness instead of fear and loathing. That to me is so important and is such a huge step forward. It gives me hope for the future and a resolve to find a suitable out of home option for Lachlan where we can still play an active role in his life.

I will finish with one of Tim's deep thoughts. Telling me:-

"having SMS is like having a door without a door knob, still a door, just harder to use"

Thank you to Kate for sharing your story it is greatly appreciated. AGSA.