



GSNV Update

January / February 2007

GSNV NEWS

Happy New Year!

We hope you enjoyed the festive season – or at least weren't too stressed out by it – and are feeling refreshed and ready for the New Year! GSNV has lots of New Year's Resolutions... Genetic Support Awareness Week in July...another fabulous Family Day... to keep you informed and inspired with our Newsletters and website...and lots more! Keep reading our newsletter and web site for details!

Small Grants Project

GSNV invites applications for its 2007 Small Grants Project. Amounts of up to \$400 are available for special once-off projects that benefit people with genetic conditions. The information sheet and one-page application form enclosed with this newsletter. *Applications close Friday 20th April 2007.*

GSNV Coffee Morning – Monday 16 April, 10am

GSNV invites people with genetic conditions, parents, partners, siblings, grandparents and support group reps to join us for morning coffee on Monday 16 April at Wingate Ave Community Centre in Ascot Vale. This is a great opportunity to meet others in similar situation (even if they aren't affected by the same genetic condition), network with people from Victorian support groups etc. Please see the brochure for more details, or phone Leah on (03) 8341 6315.

GSNV Workshop: Starting a Support Group – Tuesday 27th March, 7pm

Thinking about starting a support group? Just starting one? This workshop will provide practical tips and strategies for starting a support group. To find out more, or to RSVP, please see the brochure enclosed or call Leah on 8341 6315.

GSNV Family Book

Just a reminder that we are seeking photos, drawings, stories, poems, words of wisdom, or anything else that reflects who you are – and, ultimately, who we are – for our Family Book. Anthony Condell is our star contributor this month, with his story featured on page 4. If you'd like to contribute, but aren't really sure what to do, give Catherine or Leah a call on 8341 6315. We'd love to chat with you, and help you come up with ideas, or help you polish the ones you have.

GSNV CALENDAR	INSIDE THIS ISSUE
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Family Day Report

Wasn't Family Day terrific? We had a great turn out, with over 50 people (including about twenty children) meeting up at the Hawthorn Community House for a barbecue and lots of Christmas cheer. Even the weather co-operated! Best of all for Catherine and Leah was the chance to meet GSNV members in person (mostly, we only get to talk to you on the phone or by email) – some of you attending your first GSNV event! We hope you enjoyed the chance to meet up with other families, and have a fun day out with the kids.

Special thanks to our volunteer helpers, in particular Sharon and Tammy, who saved us all from disaster when both barbecues failed, by cooking up dozens of sausages, mushrooms, veggie-sausages and onions in the Community House Kitchen. Lots of fun was had out in the park with the giant bubble blowers, and inside making badges, working on a joint GSNV poster, and learning how to make paper cranes. Our face painters added colour to the day, but the highlight of the day was a visit from Santa Claus, who came bearing a sackload of gifts for adults and children alike, kindly donated to us by the elves at the Museum Shop and Cystic Fibrosis Victoria (not to mention lots of lollies and chocolate). Carly, our new President, took photos with her polaroid camera (and had photos taken of her, too – see below), and the children were fascinated to watch their photos developing as they looked at them. The day ended with a celebration cake in GSNV blue, covered with gerberas in white chocolate, and a firm resolution to do this again in 2007... Thank you to all who attended, and all who helped make this such a special day.



Australasian Genetic Alliance

The Australasian Genetic Alliance (AGA) is a network between umbrella support groups around Australia and New Zealand. The AGA represents people with genetic conditions at a national and international level, and provides an opportunity for collaboration between states and countries in the region. GSNV is a founding member of the AGA and represents Victoria. GSNV is hosting this year's AGA meeting in Williamstown on Monday 26th February. To find out more about the AGA, please contact Leah on 8431 6315.

GSNV Website

Have you visited the GSNV website recently? While the look is the same as ever, the content has undergone quite a few changes, with a new Advocacy section (including a list of Advocacy groups and Advocacy News), a section for support groups and people who would like to start one, and frequent updates with interesting sites or information. It's definitely worth a look, though we say so ourselves! Got a website of your own? Consider linking to us... and while you're at it, drop us an email and tell us about it – we'd love to list as many of our members' websites as possible.

GSNV Pens and Pins are still on sale from the office – visit the website at www.gsnv.org.au, or call the office on 8341 6315 to order yours today!

GROUP EVENTS

New Zealand PKU Camp 2007 –23-25 February

Venue: Epworth Retreat and Recreation Centre. Email cmcmahon@adhb.gov.nz for further information.

Australian Leukodystrophy Support Group AGM – Sunday, 25th February

Time: 1:30pm-3:30pm; Venue: Ella Latham Meeting Room, 1st Floor, Royal Children's Hospital. RSVP: by 15 Feb to 9584 7070 or mail@alds.org.au

Ménière's Support Group of Victoria Seminar – Sunday, 1st April

Time: 2pm-4pm. Venue: Deaf Children Australia – Auditorium, cnr St Kilda Rd and High St, Prahran. Seminar title: "Building Defences - Natural Health Strategies" Speaker: Dr Noone; RSVP: by 30 March to 9783 9233 or info@menieres.org.au .

OzED Family Day (Victoria) – Sunday, 6th May

Time: 11am-3pm; Venue: White Hills Botanical Gardens (Bendigo) – Epsom Road, White Hills. RSVP: info@ozed.org.au.

CONFERENCES

2007 Biennial Australian Conference on Autism Spectrum Disorders – "Creative Futures" – Gold Coast, 14-16 March

Venue: Royal Pines Resort, Gold Coast, Queensland. Further information: <http://www.astmanagement.com.au/autism7/>

4th International Congress on Shwachman-Diamond Syndrome – Boston MA, 10-12 June

Venue: Conference Center at Harvard Medical School. Keynote speaker: Our keynote speaker is Dr David Nathan. Further information: www.dsdcongress07.com .

GROUPS AND SERVICES

XXWhy: The Australian Support Group for Klinefelter Syndrome * NEW GROUP*

James has recently started an online support group for people with Klinefelter Syndrome. Further information: <http://www.xxwhy.com> or James@xxwhy.com.

Jeune Syndrome Association * NEW GROUP *

Claudine has started the Australian Jeune Syndrome Association, and invites people to join. Further information: <http://home.vicnet.net.au/~jeunesyn/> .

Smith-Magenis Syndrome Support Group

Smith-Magenis Syndrome Australia provides an opportunity to speak with other people in similar situations. Contact: Mr Bernie Benson

(secretary); 0407 920 881 (BH), (02) 6926 1612 (AH); benson@dragnet.com.au

Supporting Aussie Kids with Kabuki Syndrome (SAKKS)

SAKKS is an Australia-wide group with representatives and contact people in each State. Further information: www.sakks.org.

Support After Fetal Diagnosis of Abnormality

Support After Fetal Diagnosis of Abnormality is a facilitated shared experience group for parents who have ended a pregnancy because of fetal abnormality. Contact Alison Thornton on 9344 2121 for more information.

Full details of these events and services can be found at <http://www.gsnv.org.au/pages/news&events/news.php>. If you don't have access to the internet, the GSN(V) staff are happy to print the information and post it to you. Please call us on 8341 6315. For more information on anything you've read here, or to have your group's event advertised in the GSN(V) Update, please phone Leah or Catherine on (03) 8341-6315 or e-mail info@gsnv.org.au .

PERSONAL STORY

What am I? Anthony the Sissy? – by Anthony Condell

I have Ehlers-Danlos Syndrome, my skin is silky and smooth more than any cosmetic company can ever imagine and it's delicate just to make it more annoying. My ligaments don't hold me in the right place that much and my scars are all there to show evidence of almost every fall I've ever had.

I'm truly blessed because I look pretty normal and I was generous enough to share this stupid gene malfunction with my two wonderful daughters.

I recall being in grade 2 and the head master came in with his shorts and white socks and explained how much fun school camp would be. I put up my hand and told him I didn't think I would be able to go because I would hurt myself, he in turn told me I should untie those apron strings and not be a sissy, get some football into ya! In prep a teacher told me not worry that I'd been kicked, never mind the gaping bleeding wound on my shin!

But I also recall the caring grade 5 teacher who took me to hospital when her morning exercise program went haywire as I clumsily jogged into a park bench and ripped a huge hole in my leg.

In High school some older girl pulled a towel from under my arm, I turned still hanging on to the towel, my knee cap popped out I was crying, but I could hear her freaking out just as much!

In high school my classmates complained in disbelief when a team captain who didn't know me selected me first from a line-up to join his team because I looked capable, in retrospect my classmates were right I was bad at sport. (Cmon you guys I tried twice as hard as you ever did.)

Once I broke my wrist and the doctors wrapped it twice, I'll bet they wrapped it too hard and my

hand when blue so they cut the cast off and they cut my arm! They then stuck a couple of strips on the cut and six weeks later they took off the plaster cast and my mum and I discover this gaping cut on my arm! We get ushered into this office where apologies are offered, I remember green leather and nice furniture. Doctors and nurses always wrap me too tight, every time I've had to unwrap bandages when I get home, you can't make them understand how soft EDS skin is. "The bandages have to be firm" they'll say, ...yeah let me wrap you with equivalent force!

I bring my kids to the RCH when I have to and all these childhood memories flash in my mind, mostly good feelings. My wife has a hard time dealing with our delicate kiddies. It's almost like crossing into EDS world and feeling helpless, probably like most parents of special kids. Never mind - we got heaps of love and hugs, actually we're running out of 44 gallon drums to put it all in.

Well that's my little whingey story, my elbows are sore from leaning on this desk and I just bumped my knee on this stupid computer desk, but I am yet again reminded to soldier on and not be a cry baby. Now we have miracle lotions in our house - vitamin E oil, and that blue clear cream for bruises and vitamin C, we are ready for the world, bring it on!

Anthony's story will appear in the GSNV Family Book. We thank Anthony for giving his permission to publish it in this newsletter – we don't think he's a sissy at all (Real Men DO write stories for the Family Book)! Keep sending us your stories – they are the best part of our newsletter, and we can't do them without you!!

CONNECTING FAMILIES

Ehlers Danlos Syndrome

Debbie is looking for other families affected by this condition, with a view to starting a support group for Ehlers Danlos in Australia.

If you are interested in being part of such a group, please contact her on 0427 797 885.

Beckwith Weidemann Syndrome

Monique has a daughter affected by Beckwith Weidemann Syndrome and would love to

hear from other families affected by this condition. To contact Monique, please phone 0401 869 392.

GENETIC TESTING

Genetic Health Services Victoria - Genetic Testing Update

Following a year of Genetic Health Services Victoria (GHSV) documenting and recording all requests for unfunded genetic testing, a report was compiled by GHSV clinical director, Professor Agnes Bankier, and presented to DHS requesting additional funding to support these requests. DHS has responded by allocating GHSV additional funding for genetic testing for the individuals and families who use our services.

Of course this is not an unlimited pot of money, and so a system has been put in place where requests will be triaged on the basis of genetic risk and clinical

management. This is to make sure that the money is utilized in an equitable way, and so that we do not run out of funds in the first few months of the year. To date this has been a successful process and GHSV has been able to fund a number of requests for tests which have previously been unfunded, particularly in the area of prenatal testing.

If you have any further questions regarding this funding please feel free to contact your genetic counsellor/ clinical geneticist or Genetic Health Services on 8341 6201.

- Sue Fawcett, Genetic Counsellor and GSNV Committee Member

GRANTS AND FUNDING

William Buckland Foundation – ANZ Charitable Trusts

The William Buckland Foundation strives to identify and support initiatives that can make a lasting, significant and positive impact on the well-being of the Victorian community. The Foundation will support innovation in practice; strengthening the ability of not-for-profit bodies to deliver effective services; and building knowledge within the not-for-profit sector and the wider community. Grants are generally awarded for the period of one year, and are usually for a minimum of \$20,000. Emphasis is on

the prevention of problems, addressing the causes of disadvantage; response to an identified area of need in the community; making a significant contribution to the body of knowledge of an area; or other positive impact on community wellbeing, especially for disadvantaged groups and rural communities. Closing dates for 2007 are 1 January, 1 April, 1 July and 1 October. Call the ANZ Charitable Trustees on 1800 808 910 for details, or visit <http://www.anz.com/aus/fin/Trustees/guNamed.asp#WillBuck> to download a copy of the application form.

RESOURCES

Green PC opens 'Super Store'

Green PCs are recycled personal computers that are refurbished and repackaged into usable Internet ready computers and then made available to low-income communities, individuals and community organisations. They have now opened a "Super Store" in the

Brotherhood of St Lawrence shop at 462 Geelong Rd Footscray, with a wide range of PCs on display, as well as 2nd hand printers and other peripheral devices. You can visit Green PC online at <http://www.greenpc.com.au> or call 1300 306 645 for more information.

MEET OUR MEMBERS

Trichothiodystrophy (TTD) Support Network

"The TTD Support Network is a place for families as well as patients who are looking for information, contact and support for people living and dealing with trichothiodystrophy... We are not doctors or specialists, we are concerned parents who want to provide the best we can for our children"

About the TTD Support Network

The TTD Support Network brings together families of children with TTD from around the world. This group is founded by parents of children living with this disorder and would like to share and support other families living with the same. As well, it provides helpful information to those who provide care or interact with any persons facing this disorder.

The web site is hosted by Maria, whose son has TTD. It contains her story, and a link to her blog, where we get a glimpse of the day to day life of a child with TTD. The site also contains a factsheet on TTD, links to the online support group, media, and further reading on TTD research.

The TTD Network has a strong focus on raising awareness in the medical community

and among the general public. As a result, children with are now getting diagnosed a lot earlier, allowing parents to minimise their UV exposure.

History of the TTD Support Network

The TTD Support Group was started by Maria Liistro in August, 2002 after her son, Sammy-Joe, was diagnosed with TTD at the age of 13. Maria searched the net for answers, and found other parents after posting a plea on the internet and setting up a webpage. The group has been a huge success, and members feel like 'one big worldwide family' – one with 16 children in Australia and 57 other children scattered across the globe! The group stays in contact through an online messageboard, but hopes that one day face-to-face meetings will be possible.

About Trichothiodystrophy

Trichothiodystrophy is a rare recessive DNA repair disorder, in which the cells are constantly breaking down due to UV exposure and cannot be repaired. It is a seriously disabling disorder, with a severe skin affliction and serious developmental defects and growth retardation. Children with TTD are frequently sensitive to light and UV, and experience extreme pain and blistering, as well as fatigue, dizziness, headaches and other physical problems when exposed to sunlight.

Diagnosis

Diagnosis is made by studying the hair mounts, and by amino acid analysis which demonstrates decreased high sulphur matrix proteins. The hair breaks and fractures once it emerges from the skin and is exposed to the environment, and resulting in brittle, short and sparse hair.

Treatment

There is currently no treatment for Trichothiodystrophy.

Contact the TTD Support Network

Address: 13 Hursley Court, Craigieburn VIC 3064

Email: sammyjo2@bigpond.com

Websites: <http://www.vp-it.com.au/sammyjoe/index.htm>

<http://www.caringbridge.org/ok/sammyjoe>

Support group: <http://health.groups.yahoo.com/group/TTD-support/>

10th Floor, Royal Children's Hospital, Flemington Road (PO Box 1100), Parkville VIC 3052

Ph: (03) 8341-6315 Fax: (03) 8341-6390 E-mail: info@gsnv.org.au

Trichothiodystrophy: a mother's story

My son Sammy-Joe has TTD and has only recently been diagnosed. He is now 17 years old. He was born by emergency caesarean section at 38 weeks. He was smaller than expected for a child at that age, and his head was 26% smaller compared to the norm. He was born with curly, brittle hair, short and sparse, small eyes, dry skin and very floppy. He weighed 5lb 2oz and lost a dramatic amount of weight in the first two days of his life.

He lost all of his hair after having a temperature, while having a bath in the hospital. He had difficulties suckling, and failure to thrive. Because of his poor immunity, he has had many stays in hospital throughout his life undergoing extensive tests, Gammaglobulin treatment, surgery and rehydration. He has lactose intolerance, ataxia, speech delay, autistic characteristics, poor weight gain and stunted growth. Certain sounds and lights affect him, triggering him to scream as if in pain, which I now believe at times truly hurts him.

In our world it is easy to understand things, but in Sammy-Joe's world it is challenging. His condition still challenges him in many ways, and gives him difficulties that other people probably wouldn't be faced with and wouldn't need to worry about every single day - things like worries about light, things like blisters constant head pain and severe burning feeling on his skin just from getting undressed to put his other clothes on. It has taught him tolerance, patience and most importantly understanding. I always say that regular people wake up and start a new day leaving all their worries from the days that have passed behind them - but not Sammy-Joe. He wakes up and his worries are still there and this gives him great difficulty in understanding why? He doesn't understand why he has to be sick, and says out loud "help me, the lights hurt me, the sun hurts me help me."

Aside from all the trials and battles in his life, he is a happy child that has a passion for books, videos, nursery rhymes, The Muppets, animals, stuffed toys and James Hird from the Bombers. My sister in-law once said, "imagine if everyone had the same passion for things, or loved the same way Sammy-Joe does. This world would be a better place".

I have often been asked what does the future hold for my son's life, and I would be lying if I

said it doesn't scare me. In fact it scares me so much my response is always "nobody really knows". The other children with TTD that I am in contact with are all younger than him, and because this disorder is so rare, I have only been able to find 16 other children in Australia, 73 world-wide.

In 2003 we travelled to America to meet up with scientists and doctors from NIH and MAYO Clinic and UCSF Hospitals which helped change the quality of his life. Although not cured his quality has greatly changed.

Sammy-Joe is my pride and joy, my heart and soul, and my whole reason for living. He is truly a miracle child and a survivor, an inspiration to his family and friends. His brother Christian adores him, and he is the apple of his Dad's eye. We love both our children and have taught them to be very proud and stand tall, despite all the challenges that life brings us.

Sammy-Joe constantly amazes me with his incredible happiness and his sheer love of life. Like all parents, we want everything for our son, and thinking ahead about what the future may, or may not hold, makes me strengthen my determination to see that my child has every opportunity and experience he deserves.

Sammy-Joe's diagnosis has forced us to re-evaluate our entire lives and the way we live. We surround ourselves with family and friends who understand and are sensitive to our situation. We decided as a family that whilst this diagnosis is devastating and we have no control over it and the final outcome, we do have control over the way we manage our situation and the way we choose to give Sammy-Joe a quality of life that best suits him. The simple things in life really do mean much more, and spending quality time with Sammy-Joe means so much more to all of us. We all live day by day and enjoy every moment with our precious little boy.

I'd like to tell you all that as long as we are blessed with Sammy-Joe, we will have all that we need. Everything else seems so unimportant. He will always touch hearts, and even change lives with his smile.

More of Sammy-Joe's story can be found on the TTD website. Thanks, Maria and Sammy-Joe, for sharing your story.

CURRENT EVENTS

UN General Assembly Adopts Groundbreaking Convention on Rights of Persons with Disabilities

On December 13, the UN General Assembly adopted the first new human rights treaty of the twenty-first century marking the culmination of nearly two decades of work on protecting and promoting the rights of persons with disabilities and a major shift in the way the world treats its 650 million disabled people.

The Convention was the most rapidly elaborated instrument ever, accomplished in just three years because of the dedication of its supporters, including a large segment of civil society who had lobbied heavily both in person and over the Internet, including with Governments.

Don MacKay (New Zealand), Chairman of the Ad Hoc Committee on the Protection and Promotion of the Rights and Dignity of Persons with Disabilities, said that, theoretically there was no need for a new Convention as existing human rights instruments already applied to persons with disabilities in the same way they applied to everyone else. Unfortunately, however, that was not the reality. The Convention was a "benchmark for future standards and action". The key would be effective implementation, which required coordinated action by disability organizations, cooperation among States and the mainstreaming of disabilities issues into development assistance programmes.

All delegations agreed that the Convention's adoption ushered in an important paradigm shift towards recognizing disabled persons as rights holders and active members of society rather than objects of charity.

The Convention on the Rights of Person with Disabilities is founded on the principles of:

- (a) Respect for inherent dignity, individual autonomy including the freedom to make one's own choices, and independence of persons;
- (b) Non-discrimination;
- (c) Full and effective participation and inclusion in society;
- (d) Respect for difference and acceptance of disability as part of human diversity and humanity;
- (e) Equality of opportunity;
- (f) Accessibility;
- (g) Equality between men and women;
- (h) Respect for the evolving capacities of children with disabilities and respect for the right of children with disabilities to preserve their identities.

(quoted from Article 3 of the Convention)

Further articles of the convention include accessibility, equal recognition before the law, freedom of expression and access to information, liberty and security of the person, respect of privacy, living independently and being included in the community, education, health, work and employment, and adequate living and social protection standards. The text of the convention can be found at <http://www.un.org/esa/socdev/enable>.

REQUEST FOR SUPPORT

The Burrodiera Centre – a new service for families affected by Rare Chromosome Disorders & Other Developmental Disabilities

The Burroodiera Centre is a national centre that will be located in country NSW dedicated to the quality of life and independence of those affected by rare chromosome disorders and developmental disabilities. The Centre is asking for your support by completing a survey (available from the GSNV website or by ringing Nicole on 02 6624 7128), which will help demonstrate to funding bodies the demand for such a centre, as well as assisting in planning services.

WORKING AND CARING

The Working Carers Portal – www.workingcarers.org.au

Do you work full- or part-time while caring for someone with a disability? This website may be for you. While it's based in NSW (making its services directory less relevant for Victorian readers), there's lots of information on self-care, money matters, negotiating with employers, working with support services and all sorts of other goodies – as well as a dictionary of medical terms, personal stories, a newsletter, and information for employers. If you're online, it's certainly worth a look.

Extract from 'Fact Sheet for Employers: Carers and Work'

Quoted with permission from the Working Carers Support Gateway, <http://www.workingcarers.org.au>

The population of employees with care responsibilities has grown, and will continue to do so, due to a number of factors. These include the ageing of the population, the growing preference of women to be in paid work, and government policies that support people to live in the community. Employers can respond to these trends by supporting employees with caring responsibilities.

Juggling the competing demands of work and caring can be stressful for employees, but there is good evidence that the right supports in the workplace can make a real difference for the employee, and have benefits for the employer as well.

For employers, the benefits of supporting carers include:

- Reduced costs through less employee turnover, lower absenteeism and sickness, and higher rates of return on investment in trained and experienced employees;
- Improved labour flexibility through the availability of a larger, more diverse labour pool, improved access to scarce skills, and a better ability to meet peaks in workload;
- Enhanced motivation of workers, including improved morale, greater staff loyalty and reduced levels of employee stress.
- Improved business performance; and
- Good corporate citizenship and enhanced corporate image.

Every caring situation is different. The kinds of assistance that carers provide to their family member will vary depending on factors such as the age of the person receiving care, the nature of their disability or illness, the length of time they have needed support, whether or not they live in the same household, cultural background and the community supports that they receive.

Carers' needs may be very simple, such as the ability to leave work on time, or access to a telephone so that they can check on the person they support. Some carers will benefit from ongoing flexible arrangements, while other will need them on a temporary or sporadic basis. Also, people's circumstances and needs may change over time.

Research project - Questionnaire for parents/carers who are working full-time & caring for a child with chronic illness/disability.

Are you working full-time and caring for a disabled or chronically ill child? If so, your participation in a University of Western Sydney national survey would be appreciated. Its aim is to identify your needs and provide valuable information to plan better support services for you.

Your participation will involve completing a questionnaire and returning it in a reply-paid envelope that will be provided. It will take about 25-40 minutes of your time and your confidentiality and anonymity will be maintained. Questionnaires can be obtained from **Ajesh George** on **02 9845 0422**, or completed on-line at www.workingandcaring.org. For further information about this study, please call: Professor Lesley Wilkes on 02 4734 3181, Email: mcmanua@wahs.nsw.gov.au

Got any news for this page? Know of an advocacy group or organisation that should be featured? Contact the office on 8341 6315 or email info@gsnv.org.au to tell us about it..