



# FRAGILE X TRUST (NZ) NEWSLETTER

June 2008

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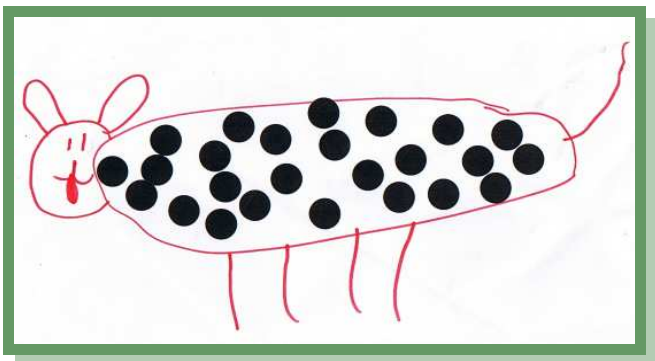
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### Special points of interest:

- US Fragile X Conference
- Fragile X Awareness Day
- Fragile X in Australia
- Two Family Gatherings
- Support the Fragile X Trust

Thanks to the generous support of the Minister's Discretionary Fund and the US National Fragile X Foundation, The Fragile X Trust is able to assist six parents attend the 11<sup>th</sup> international conference on fragile X syndrome, which will be held in St Louis, Missouri, this July.

The attendees include Anita Nicholls, Judith Spier, Seniorita Laukau, Jodi Heenan and Anne and Niel Smith and their two children. Anita and Jodi have been invited to make a presentation on their highly successful education programme: No Longer Fragile (see December 2007 newsletter). This will be only the second time that New Zealand has been represented at the conference (Anita attended the last conference) and, with Seniorita attending, the first time that a Pacific Island has been represented.



**Dalmatian** by Danielle Spier, age 11

## Cheers from the Chair



Greetings to the New Zealand fragile X community and to our friends overseas.

Apologies for being so late with our first newsletter of 2008. It's been a very busy year so far for our committee with numerous awareness- and fund-raising activities and the roll-out of our new education program. The January issue of "North and South" magazine featured a five page story about fragile X that was based on our own family's experience. Feedback on the article was very positive with many of you passing the article onto family, friends and colleagues. The article also resulted in several enquiries about fragile X and certainly increased calls to our new toll-free number. This service is proving to be a huge success with calls from new families now becoming a regular event.

Chris and I recently had a brief holiday in Australia and we took the opportunity to attend a committee meeting of the Fragile X Association of Australia in Sydney. It was a very worthwhile visit and we plan to have better links with our Australian colleagues, sharing ideas and information while working together on specific issues, such as newborn screening, and speaking tours by international fragile X experts. There are many more fragile X families and specialist services in Australia than here, so to get information on a topic that concerns your family we can help to connect you with families and professionals in Australia. For example, for those of you interested in PGD see the letter we have reproduced from the Fragile X Association's newsletter.

In July, Judith Spier, Jodi Heenan, Senorita Laukau and I will fly to USA where we will tour the MIND Institute in Sacramento with Louise Gane and talk to Randi Hagerman's team. We will then travel to St Louis to attend the International Fragile X Conference where Jodi and I will give a presentation on our new education programme. Anne and Niel Smith will also attend the conference with their two children, taking the opportunity while in the US to have their son assessed by Randi's team. The six of us are determined to make the most of the opportunity and bring back a wealth of information to share with local families. All are making a significant financial contribution to represent NZ at the conference and we are very grateful for sponsorship from the Minister's Discretionary Fund and the US National Fragile X Foundation.

There will be opportunities at the Mind Institute and the conference to ask questions on your behalf. Please contact me if you have questions. We will also be able to bring back books and resources. So, if you have seen anything that is only available in the US get your request to me as soon as possible.

The end of the year will be a busy time as the Trust will have display booths at the NZ Paediatrics Society Conference in the Bay of Islands and the Autism Conference in Christchurch. We will also be holding two family gatherings - one in Wanganui and one in Christchurch (see next page). I hope to see you there!

*Anita*

Call me toll-free 0508 938 0552

## Website changes

### [www.fragilex.org.nz](http://www.fragilex.org.nz) (library)

Darryl and Tracey have updated the library catalogue and soon we hope to be able to process borrower's request with a online form.

#### **Library loan conditions:**

- χ There is no limit to the number of items that can be borrowed at one time.
- χ We cover the costs of delivery to the borrower.
- χ The borrower is required to pay the costs of returning borrowed materials to the library
- χ Loans are for one month unless special arrangements are made.
- χ Because this is a free service, we request that all care is taken to ensure that loaned material is returned in good condition.
- χ The Fragile X Trust (NZ) does not necessarily endorse the contents of all the publications in our library.

#### **Return loans to:**

Tracey Cockcroft , Fragile X Trust Librarian  
2 Trigg Rd, Huapai, Auckland

## *Fragile X Awareness Day*

Tuesday 22 July is International Fragile X Awareness Day. Numerous activities are being organised throughout the world for the week starting 20 July to raise awareness about fragile X and to raise funds for local support groups, and research and treatment programmes. This week also coincides with the international fragile X conference in St Louis, Missouri.

What can you do to raise awareness about fragile X in your community?

- χ a fund-raising activity
- χ a talk to a local community group
- χ distribute pamphlets to medical clinics, schools and kindergartens
- χ write a letter or article for your local paper

If you'd like to do something, contact us for pamphlets and assistance.

# Fragile X Across the Ditch



Chris Hollis, Jocelyn Sellwood, John Kelleher, Anita Nicholls, Doug Rodgers and Kathryn Murphy

Last month Anita and Chris had the opportunity to combine a holiday with a visit to the headquarters of the Fragile X Association of Australia. It was a huge thrill to (a) successfully negotiate the Sydney traffic and arrive at the Kelleher's stunning home in Balgowlah Heights on time and (b) to meet the dynamic committee that is driving the association's activities.

It was a particularly exciting committee meeting because it was the inaugural meeting of the new national organisation. In contrast to New Zealand, which has had a national fragile X organisation for over a decade, Australian states have tended to operate separate support networks. Under the leadership of president John Kelleher and his able team, the association will now incorporate support groups from New South Wales, Western Australia, Victoria and the soon to be formed Queensland organisation. It is to be hoped that in the future South Australia, Tasmania and the Territories will also be represented.

The Fragile X Association of Australia funds several research projects and also funds clinics in Sydney and Melbourne. For more information on their services and activities and to view their newsletters visit their website: [www.fragilex.org](http://www.fragilex.org)

Jocelyn Sellwood runs the Sydney-based office and if you have family or friends in Australia who wish to join or support the national organisation or regional support groups, please email [fragxadm@bigpond.net.au](mailto:fragxadm@bigpond.net.au) or phone 1300 FX INFO (1300 394 636 Tues/Wed/Thurs)

***NOTE: The letter from Nicola Jones on the next page (page 5) is reprinted with permission from the June 2008 issue of the Fragile X Association of Australia newsletter "fx info". Please contact the Fragile X Trust (NZ) if you'd like to be put in touch with Nicola***

## PGD – an Australian family's experience

On the 15th of June, Rick and I became the proud parents of twins. Dominic Andrew and Bronte Anne were born at 31.3 weeks, quite unexpectedly, weighing 2kg and 1600g respectively. They are at home with us now after spending a week in Royal Nth Shores' high dependency unit and 4 weeks at the San's special care nursery. Both are doing well and stacking on the weight. Dominic will need a small operation on a hernia in a couple of weeks but apart from that we are only suffering due to sleep deprivation and the hardest work we have ever had to do. Rick and I have always wanted lots of kids. When Maddie was born and we got the diagnosis of FXS, we thought that Maddie may be our only one. Fate intervened and we found we were pregnant with Jordan. Through CVS testing, Jordan was found to be FX free.

At every Association AGM, we have some very good speakers and a couple of years ago, Cynthia Roberts, one of our members and Scientific Director with Sydney IVF, gave a very good talk on PGD IVF. I am not going to go into the science of this procedure but it stands for Pre-implantation Genetic Diagnosis In Vitro Fertilisation. Quite a mouthful! Anyway, it involves the whole IVF procedure to grow embryos and before they are implanted, around 4 to 6 cells are removed and tested for Fragile X Syndrome. Only FX free embryos are implanted. Cynthia also explained that with Medicare rebates, the Medicare safety net and private health insurance, the cost isn't really that great.

We decided to give it a try. We had meetings with a nurse, a scientist, an obstetrician and a counsellor, who all explained their roles in the procedures and made sure we were properly prepared. The thing that bothered me the most were the needles. It was to be at least one a day, sometimes two and maybe three. The injections are hormones that stimulate egg production and ovulation and of course, the injections for the blood tests. I must admit that I was a little uncomfortable as well as emotional through the processes but I did want to have another child. Rick became the nurse and was in charge of giving the needles. We did try two stimulated cycles to only get 4 viable, FX free embryos. This, and the subsequent transfer of embryos lasted around a year. The first 3 embryos were transferred one at a time over a period of 10 months and were unsuccessful. The fourth, and only girl, was the last one to be tried. We had already decided that if this one didn't work then we wouldn't try again. It was successful.

Those of you who have followed the story are now thinking if one embryo was put in, how did you get twins? Well, we went for an ultrasound at 7 weeks and discovered we were having twins, and after consulting our obstetrician we believed the embryo had split on transfer, therefore resulting in identical girls. We were stunned but thrilled all the same. The next test was a CVS to ensure there was no FX. We were quite confident that everything would be fine because the embryos had been screened but we wanted to be sure. The results came back with two pregnancies one male and one female. How did that happen? Surprise! One IVF and one natural baby, both without Fragile X and due 14th August. Well, all was going well until 11pm on the 13th June. The water broke on one baby and I was rushed to hospital where they held back labour until 4am on Friday the 15th June. The babies arrived and were very well looked after by the neonatal doctors and nurses of RNS until their transfer to The San where we were again spoiled by the dedicated staff.

IVF and PGD can pose some ethical questions and I realise that some people may not agree with the whole process but for us it has been a very positive yet emotional experience. If anyone is considering it or would like to speak to me about it, I am available on the parent support line number. However, please have patience with me as I now have 4 kids, 1 with FX, 1 with real attitude and very young twins but I do love every minute of it, well, nearly every minute of it.

*Nicola Jones*

## Family News

Hi one and all. It has sure been a while since I have put anything in the newsletter. So here goes.

Craig is now 27 years old and is an adult now. When we first started out getting the diagnoses of Fragile X and also when we found out Craig was going blind, trying to find the right therapies and education and list goes on. Life back then was hard enough. Fighting the system for everything, which by the way never changes. When Craig left school that's when you realize there is not a hell a lot out there. You really have to look around. We made some balls up along the way but learnt real fast. After a lot of kicking, screaming and letter writing to parliament we finally got Craig's needs meet. It took around 4 years to finally get someone to actually listen to us and realize we weren't asking for the moon. Kevin & I thought yes things will run smoothly after Craig left school how wrong we were. The BS one has go through to get right placement is unbelievable. We wouldn't wish that on anyone. So before your child/ children leave school be very proactive and make sure the Powers at Be so to speak actually listen to what the hell you are saying or else you be pushing it up hill.



Craig is doing very well. He has a full and active life which has a purpose. Craig is still working at the SPCA taking care of the Rabbits which he has been doing for 4 years now. He also works at the Mangere City Library and at Ambury Regional Park planting native seedlings. Craig also does other things with Focus 2000. Craig is fully supportive in all the things he does.

*Vicki Williams*

Kevin & Craig while we were on holiday on Norfolk Island in May

Hello from Dunedin!

We had the privilege of participating in the South Island Independence Games for 2008 in April. It was a fantastic event- it is for ages 8- 16. There were approximately 100 competitors. All the children win a gold medal – they really are all champions with so much effort going in. Adrian won the prize for the best Mascot (the Otago Highlander) - Ruth Dyson who handed out the prizes thought he was the real one!

I hope you are all keeping warm this winter.

*Kim and Adrian Caffell*



# ANXIETY – Just how bad is it?

Marcia Braden PhD



Many clinicians who see individuals with FXS hear from parents on a daily basis how anxiety affects their child's behavior. It is not uncommon for a child to

throw a tantrum before going on a trip to a favorite play spot, activity or recreational facility. Adolescents and Adults may retreat to a bedroom and refuse to come out. It is hard to understand how these kinds of events can trigger such a negative reaction.

Like many things about individuals with FXS, this scenario doesn't fit a logical pattern. When an activity or experience has been fun, one would expect the child to be excited and recall positive feelings about the experience. There are several important factors to consider in order to better understand this phenomenon.

A person with FXS has difficulty modulating incoming stimuli. We know from a variety of research venues, that too much sensory input and a pervasive discomfort from excitation can promote hyperarousal resulting in "behavioral meltdowns". The mere fact that the child enjoyed the activity at another time isn't enough to override the initial feeling of being overwhelmed.

Anxiety is usually accompanied by physical symptoms such as a racing heart, blushing (red ears or neck), sweating and nausea. Experiencing those physical changes can also create more fear, followed by panic. The person with FXS may also be impacted by an executive function deficit that interferes with his ability to remember the past experience in a way that would provide reassurance and motivation to try again. When confronted with the excitement, the person with FXS may first become anxious followed by an inability to regulate his arousal level and properly manage his behavior. An attempt to avoid the situation may occur in order to endure the anticipated stress. This cycle feeds the pathology causing the behavior to escalate. It is counter therapeutic to avoid these family outings and recreational experiences, even though it is at times very tempting. In order for the child to become desensitized, he must experience

repeated exposure to the event. This takes a lot of patience with the understanding that if the time and energy is spent early, it will become less difficult and disruptive later.

There are a number of ways parents and caregivers can prepare the child for the activity. Some parents report reading a bedtime story the night before with pictures taken of a fun experience. If the child has a tendency to obsess and worry about the future, it may be better to discuss it right before leaving with time built in to employ a sensory menu. The preparation time includes utilization of calming strategies and an emergency kit of chewing gum, water bottles, audio tapes, fidget toys and other self calming supplies to take to use on the way to the activity.

It is well known that anxiety can have biological roots. Fearfulness is associated with irregularities in neurotransmitters such as dopamine and serotonin. Studies in the general population show that high levels of the stress hormone cortisol releases when one is anxious. Belser and Sudhalter have also researched the affect of arousal on individuals with FXS with similar results.

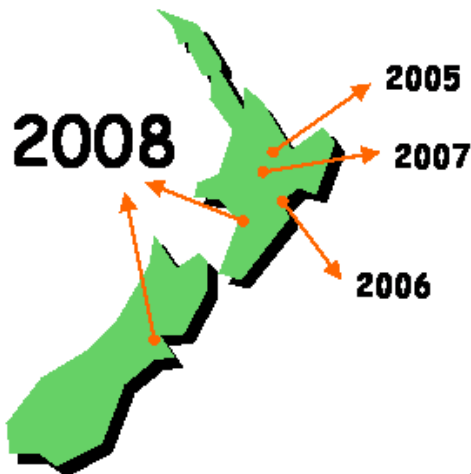
Anxiety can have far reaching effects on the life of one with FXS. Each experience can virtually shut down adaptive behavior. The fear can be so intense that the individual with FXS may revert to a primal reaction of flight or fight and become unable to access an appropriate behavioral response. The best remedy to all of this is the gift of time.

Building in enough preparation time to allow for a sensory diet, behavioral story and use of the emergency kit can slow down the process and allow a "slow motion" effect to take hold. This will also give the parents and caregivers sufficient time to react in a calm and supportive way, adding less stress to the mix.

Marcia Braden is a licensed psychologist with a clinical practice specializing in children and adolescents. In addition to her PhD, she holds a masters degree in learning disabilities and emotional disturbances.



# Two Family Gatherings for 2008!!



Due to popular demand the Fragile X Trust is heading south and organising our first family gathering in the South Island. But we can't abandon you northerners so we'll have a North Island gathering too.

We hope to be able to offer all families accommodation subsidies and greatly discounted activity fees but won't know how much we can offer until numbers are confirmed. So please let us know if you'd like to attend one or both of these gathering by **29 August**.

## Wanganui

**31 October – 2 November**  
**Whanganui River Top 10 Holiday Park**  
**460 Somme Parade, Upper Aramoho**

The programme will begin with a seminar by Louise Gane on Friday afternoon. On Saturday morning, we will take a trip up the Whanganui River in the Waimarie Paddle Steamer. In the afternoon, there will be activities around the holiday park, followed by a BBQ dinner. For those interested in participating in the Fragile X Trust's activities, our AGM will also be held in the afternoon.

**Make your own accommodation bookings as soon as possible but be sure you tell them you are part of the Fragile X Group. Phone 0800 272 664 or Email [wriver10@xtra.co.nz](mailto:wriver10@xtra.co.nz). Website: [www.wriver10.co.nz](http://www.wriver10.co.nz)**

## Christchurch

**7 – 9 November**  
**Christchurch Top 10 Holiday Park**  
**Meadow Park, 39 Meadow Street, Papanui**

The programme will begin with a seminar by Louise Gane on Friday afternoon. On Saturday morning, we will visit Orana Wildlife Park. Saturday afternoon activities have yet to be confirmed, but we will wrap up the day with a BBQ dinner.

**Make your own accommodation bookings as soon as possible but be sure you tell them you are part of the Fragile X Group. Phone 0800 396 323 or Email [meadowpark@xtra.co.nz](mailto:meadowpark@xtra.co.nz). Website: [www.christchurchtop10.co.nz](http://www.christchurchtop10.co.nz)**

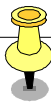
### More on the seminars and Louise Gane:

Louise Gane is an expatriate New Zealander who works as a genetic associate at the MIND Institute, University of California at Davis, the leading international centre for fragile X research and treatment. She works closely with Drs Randi and Paul Hagerman, widely regarded as the foremost authorities on fragile X syndrome. Louise is a passionate speaker and has a vast wealth of knowledge on fragile X. She will be participating in the weekend's activities and is always very willing to meet and counsel new families.

For the Wanganui seminar, Louise will build on information presented in Napier and Taupo and provide information on the latest developments on understanding and treating fragile X. For the Christchurch seminar, Louise will provide a general introduction to fragile X with a focus on behaviour and education.

**REGISTER FOR FRIDAY SEMINARS AND WEEKEND ACTIVITIES BY 29 AUGUST:  
 Phone 0508 938 052 or email [fragilex.info@nzord.org.nz](mailto:fragilex.info@nzord.org.nz)**

# Support the Fragile X Trust



The Fragile X Trust is expanding its activities and needs support from you, your families and your friends. Since our inception we have relied on grants from various charities and government bodies to run our activities and have requested only a nominal voluntary donation of \$10 from families who wish to receive our newsletters. This will continue but we ask that you consider increasing your annual contribution to the Trust. We also ask that you invite your friends and families to consider making one-off or annual donations to the Trust.

This funding will help us continue to provide the following services:

- χ **Free advice and support** to all New Zealand fragile X families and their support teams through our website and toll-free helpline
- χ **Regular newsletters and email updates** to those on our mailing and email lists
- χ **Access to a well-stocked up-to-date library** of books and audiovisual materials on fragile X and related topics
- χ **Commissioned presentations of "No Longer Fragile"**, our highly successful education programme, to parents at special education teams at preschools, kindergartens and schools throughout New Zealand.
- χ **Annual gatherings of fragile X families** at holiday spots around the country, including travel grants and discounted accommodation and activity fees for needy families
- χ **Regular workshops by international and national experts** on fragile X and related issues
- χ **Representation at national and international conferences** and symposia with relevance to fragile X families (e.g. Autism, Paediatrics, Human Genetics, Genetic Screening, Rare Disorders, International Fragile X Conference)

To make your annual donation, please complete the enclosed mailing list form with your name, any altered details and the amount donated and return it to the Fragile X Trust in the enclosed envelope.

Thank you for your support!!

# Contact Information

**Toll-Free: 0508 938 0552**

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## Change of Address?



If you are moving house, changing your phone number or email address, or simply wish to be removed from the Mailing List, please contact the Secretary.

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