FRAGILE X TRUST (NZ)

Supporting New Zealand families living with fragile X syndrome

FRAGILE X TRUST (NZ)

NEWSLETTER

August 2007

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NO LONGER FRAGILE:

An Education Seminar on Fragile X Syndrome

After months of planning, preparation and hard work (particularly from Anita), we have begun the process of presenting information and resources to families and schools. Initially we planned to present the information as a two day workshop/seminar, but after discussions with several schools and families we modified our approach to better suit what they wanted.

Anita has met with several teams and presented information, ideas and resources targeting the specific issues and concerns they have for the child they are working with. The feedback from these teams to date has been very positive with parents, teachers and specialists stating that the information was well presented and made a difference.

If you or your school have any concerns for your child please feel free to contact Anita or myself and we will assist in any way we can. Anita is a trained early childhood teacher and is a parent presenter for "tips for autism" and I am trained as a primary school teacher. We have both worked alongside a variety of specialists assisting our own Fragile X children and other children on the autistic spectrum for the last 10 years.

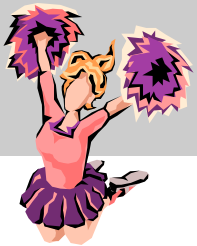
Our education seminar presents best teaching strategies for Fragile X primary school-aged children. It includes examples of resources used successfully to support Fragile X children's self-regulation and to teach reading, writing and math. The seminar has been peer reviewed by GSE staff and Marcia Braden, leading authority on educating children with Fragile X.

This programme has been developed with the generous support of the Todd Foundation. It began with two very successful workshops by Marcia Braden in Wellington and Auckland.

For more information phone Anita (04 938 0552) or Jodi (06 353 8284) or email us at: fragilex.info@nzord.org.nz

Jodi Heenan

THE TODD FOUNDATION



Cheers from the Chair

Hi to all the fragile X families around New Zealand and the professionals who work with our children. Hope you are all well and ready for the new school term. The Trust is working hard on your behalf and half way through the year is achieving the goals set at the beginning of the year.

I am just back from the Human Genetics Society of Australasia Conference in Auckland where I gave a presentation on "Family perspectives on diagnosis and testing for Fragile X Syndrome". You can read the full version of my talk in this newsletter. The main point I endeavoured to make was that early diagnosis is very important because:

- it helps families to understand why their children behave the way they do
- it helps families access behaviour strategies and interventions that make a difference
- it is important information for family planning

I also emphasised that it is important that professionals respect the knowledge and understanding parents have in relation to their own children and that testing asymptomatic siblings is appropriate when there is a strong desire within the family to do so. My talk was well received and comments made to me afterwards lead me to believe that we have achieved some improvement regarding genetic counsellors' attitude to testing siblings. We also had a fragile X booth at the conference and this provided opportunity to talk to a large number of medical professionals about fragile X.

I plan to attend the Paediatrics Association Annual Conference in November, possibly presenting as well as having a fragile X booth. I'm convinced that attending these conferences is a very effective way to get our views across to professionals.

While in Auckland I caught up with families, schools and professionals in the area and presented ideas and resources put together as part of the education seminar developed by myself and Jodi Heenan. Thus far the information and resources have been very well received with comments including "extremely useful – thank you very much", "fantastic layout and explanations", a Special Education Advisor commented "if only we had this information a year ago" and kindergarten teachers said "that was great, we would love you to come back and do a longer workshop". Over the next few weeks Jodi or I will be meeting with other families and schools in Taupo, Masterton, Hutt Valley and Levin. More information on the education seminars is in the newsletter.

Louise Gane is a confirmed speaker at the NZORD conference in the first week of November (see the ad). She will be talking about issues around screening programmes for fragile X syndrome. This will be another great opportunity to meet and talk to medical professionals and researchers about our favourite subject: fragile X. Finally, there will be a family gathering in Taupo, 27-28 October. This is the weekend before the NZORD conference and on Saturday afternoon, Louise will give a presentation on girls and carriers with fragile X. There will be a separate activity planned for the dads and children while the workshop is on. We will keep you updated with more information about the weekend as it comes to hand.

See you in Taupo
Anita, Fragile X Trust (NZ) Cheerperson

2007 Fragile X Family Gathering and Seminar

26-28 October, Taupo

Lake Taupo Top 10 Holiday Resort, 28 Centennial Drive

Join us for a weekend of fun, conversation & information

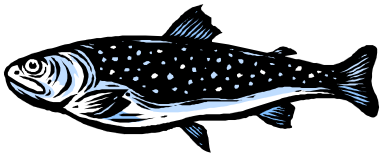
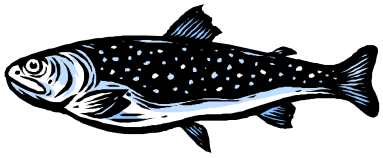
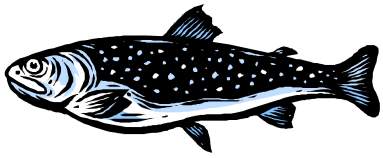
YES, they have a "jumping pillow"!
YES, Louise Gane will be there!
YES, we will go to the hot pools!
YES, some of us will go trout fishing!

Louise will present a seminar on Saturday morning (9.30 – 1.00 pm) on how fragile X affects girls and female carriers. Men are welcome at the seminar but, in lieu of child-minding services, we offer a fishing trip for dads and children.

Please book your accommodation as soon as possible directly with the Top 10 Holiday Park. Email office@taupotop10.co.nz or Phone 0800 332 121. Check out the options on their website http://www.taupotop10.co.nz

Note if they don't have the accommodation you're after, we recommend De Bretts Thermal Resort, where we will be spending much of Saturday afternoon.
Phone: 07 378 8559. Website: www.debrettsresort.co.nz

Please register with us to receive more information on the weekend's activities. Like last year, there will be a subsidised activity fee. Phone 04 938 0552 or email fragilex.info@nzord.org.nz.

Dates for your Diary

5-7 October Dyspraxia Conference, Christchurch.
The theme is "tools for learning, tools for life". Conference website: www.dyspraxia.org.nz

26-28 October Fragile X family gathering, seminar, and Fragile X Trust (NZ) AGM, Lake Taupo Top Ten Holiday Resort, Taupo. Following on from two successful family gatherings, we aim to target another holiday park with a jumping pillow for a weekend of fun, information and networking. See the ad in this newsletter.

1-2 November NZ Organisation for Rare Disorders (NZORD) Conference, Wellington. This year's conference will have a theme on genetic screening and Louise Gane is to be one of the keynote speakers. She will be talking about newborn screening for fragile X. The Fragile X Trust will be represented by Anita Nicholls and we will welcome your views on screening issues, especially carrier and newborn screening.

26-28 November Paediatric Society of New Zealand Annual Scientific Meeting, Christchurch. The theme of this meeting is "future trends" and one of the plenary speakers is Dr Hillary Cass a specialist in autism. The Fragile X Trust will be represented by Anita Nicholls at this conference. Conference website: www.paediatrics.org.nz

23-27 July 2008 11th International Fragile X Conference, St Louis, Missouri, USA. Anita Nicholls and Chris Hollis attended the last conference in 2006 and found it to be an incredible experience. The Fragile X Trust is keen to have New Zealand represented again and is looking at options for funding. If you are interested in attending this conference, please contact us: fragilex.info@nzord.org.nz

MIT locates key enzyme for reversing retardation in mice

'Elegant genetic manipulation' inhibits Fragile X symptoms

***Deborah Halber**, News Office Correspondent, 25 June 2007*
This article originally appeared on the MIT News Office website

Researchers at the Picower Institute for Learning and Memory at MIT have, for the first time, reversed symptoms of mental retardation and autism in mice.

The mice were genetically manipulated to model Fragile X Syndrome (FXS), the leading inherited cause of mental retardation and the most common genetic cause of autism. The condition, tied to a mutated X chromosome gene (FMR1), causes mild learning disabilities to severe autism.

"Our study suggests that inhibiting a certain enzyme in the brain could be an effective therapy for countering the debilitating symptoms of FXS in children, and possibly in autistic kids as well," said co-author Mansuo L. Hayashi, a former Picower Institute postdoctoral fellow currently at Merck Research Laboratories in Boston.

The study identifies a key enzyme-a chemical reaction-inducing protein-as a possible target for an FXS drug. The enzyme, called p21-activated kinase, or PAK, affects the number, size and shape of connections between neurons in the brain. Halting PAK's enzymatic activity reversed the structural abnormality of neuronal connections found in the FXS mice, said co-author Susumu Tonegawa, 1987 Nobel laureate and Picower Professor of Biology and Neuroscience. "Strikingly, PAK inhibition also restored electrical communication between neurons in the brains of the FXS mice, correcting their behavioral abnormalities in the process," he said.

There are known chemical compounds that inhibit the enzymatic activity of PAK. These compounds or versions of them may be useful in the future development of drugs for treating FXS.

"These are intriguing findings because the expression of the gene that inhibits PAK occurs in the third week after birth, which means that the neuronal abnormalities in the FXS mouse are reversed after they appear," said Eric Klann, a professor at New York University's Center for Neural Science. "This is very exciting because it suggests that PAK inhibitors could be used for therapeutic purposes to reverse already established mental impairments in FXS children."

Restoring neuronal connections

Tonegawa, Hayashi, MIT graduate student Bridget M. Dolan of the Department of Biology and colleagues study the molecules that govern the formation of neuronal connections in the brain. They explore how abnormalities in these molecules could interfere with an animal's behavior. In the brain, small protrusions called dendritic spines on the branch-like dendrites of one neuron receive chemical signals from other neurons and communicate them to the main cell body. The numbers and shapes of dendritic spines are key to normal brain function. FXS patients have higher numbers of dendritic spines in their brains, but each spine is longer and thinner, and transmits weaker electric signals, than those in non-affected individuals. When the enzymatic activity of PAK was inhibited in the FXS mice, abnormalities in their spine number and structure-as well as the weaker electrical communication between their neurons-were reversed.

Reversing behavioral symptoms

The FXS mice exhibited symptoms similar to those in FXS patients. These included hyperactivity; purposeless, repetitive movements reminiscent of autistic people; attention deficits and difficulty with learning and memory tasks.

"These behavioral abnormalities are ameliorated, partially or fully, by inhibiting the enzymatic activity of PAK," Tonegawa said. "Notably, due to an elegant genetic manipulation method employed by the Picower Institute researchers, PAK inhibition in the FXS mice did not take place until a few weeks after appearance of disease symptoms. This implies that future treatment may still be effective even after symptoms are already pronounced."

"While future studies will be necessary to further characterize the precise molecular nature of the interaction between PAK and FMR1, our findings clearly demonstrate that PAK inhibition can counteract several key cellular and behavioral symptoms of FXS," the authors noted.

In addition to Tonegawa, Hayashi and Dolan, authors include colleagues at the National Institute of Mental Health and Neurosciences; the Tata Institute of Fundamental Research in India; and Seoul National University in Korea. This work was supported by the FRAXA Foundation, the Simons Foundation, the Wellcome Trust and the National Institutes of Health.

Susumu Tonegawa and Mansuo L. Hayashi. Photo / Donna Coveney

Parents' Perspective on testing for Fragile X Syndrome

Presentation at Human Genetics Society of Australasia (HGSA) Conference, Auckland, July 2007

Anita Nicholls

When putting together this presentation I canvassed the opinions and experiences of 11 NZ fragile X families. My talk today presents their views as well as reflecting the content of conversations I have had with many other fragile X families.

Fragile X syndrome is a genetic disorder that causes a range of learning and behavioural difficulties, including intellectual disability and autism. It is caused by a trinucleotide repeat expansion on the X chromosome. If the expansion is larger than 200 repeats the gene becomes unable to make a critical protein for neural functioning. Individuals with a repeat count of 55-200 are considered carriers. An important point is that many carries have mild to moderate symptoms. Although, as a carrier myself, I can tell you we are pretty careful about what we admit to.

Despite the fact that there are many carriers in the population, fragile X is relatively rare. Just to make it more difficult for those of you responsible for diagnosis, there is a wide variation in clinical presentation. This causes a delay in diagnosis: the average age for fragile X diagnosis in the USA is 32 months (Bailey 2003). My telephone survey of New Zealand families indicates a slightly later age of 38 months.

These two handsome characters here are my two sons Ben and James. We are a pretty typical fragile X family in that we have more than one child with the condition. Ben more able than his younger brother was not diagnosed until he was five and a half yeas old. It is common that children with less classic fragile X features are diagnosed relatively late.

So is diagnosis a good thing? I put this question to families I surveyed. All said without hesitation that diagnosis was very beneficial. Reasons given were: "understanding my child"; "important information for family planning"; "knowing that I wasn't just a bad parent"; "better targeted intervention"; "informing others in the family at risk". By far the most common reason given was that diagnosis allowed the parent to understand their child. Understanding your own child's needs and reactions is essential not only to access the best intervention. It is also essential if one is to appropriately support the child through the trauma that everyday events such as supermarket shopping, visiting a friend or attending a birthday party cause fragile X children. Until diagnosis parents are unable to properly support their children.

Recently the Hagermans' team at the MIND Institute, UC Davis, have developed a newborn heel prick test. Given the benefits associated with early diagnosis, NZ families are overwhelmingly in favour of newborn screening. However families were concerned that newborn screening must be tied to comprehensive support systems that meet the particular needs of families, including genetic counselling, accurate information, early intervention services and access to family support groups.

Families have well-considered opinions about diagnosis based on their day-in day-out experience, and knowledge of what is important to members of their family. There is a feeling of ownership around diagnostic issues. As one parent said "This is our condition that only we live with". Some New Zealand families disagree with the position NZ Genetic Services takes on testing asymptomatic siblings, which is that the rights of the individual are paramount and, therefore, testing should only be carried out when the individual is 18 years or older (age of autonomous decision-making). Families believe that this late age of diagnosis denies their child the opportunity to slowly adjust to information of carrier status. One mother told me:

"It was a real shock to find out about [the fragile x diagnosis] the way we did. Fragile x is part of who you are. The earlier the information is given the better - so they grow up knowing, they can talk about it and understand it"

Siblings or close blood relatives of someone with fragile X are different than children who have no knowledge of the disorder. Siblings will ask questions such as "will I catch what my affected sibling has or will my children be affected". There will be discussions within families about the genetic implications of fragile X. Some families feel that their child would benefit from the certainty of knowing their carrier status. Allyn McConkie-Rosell presented research at the Fragile X Conference in Atlanta (2006) that found that individuals known to be at risk of carrying the fragile X gene were less anxious after knowing their carrier status compared with individuals who did not know their carrier status.

My main point is that when the ethics of a profession do not fit with the needs and beliefs of families, professionals lose the ability to communicate with and support the families they serve. Effective genetic testing and counselling needs to have an ethical framework that is inclusive of family values, belief and culture. McConkie-Rosell & Spiridirigiozzi (2004) describe just such a framework that acknowledges the importance of family perspectives.

I would like to acknowledge the support and encouragement of fragile X families, Louise Gane (MIND Institute, UC Davis), John Forman (NZ Organisation for Rare Disorders) and the NZ Lottery Grants Board.

Reference

Allyn McConkie-Rosell1 and Gail A. Spiridigliozzi1 2004. "Family Matters": A Conceptual Framework for Genetic Testing in Children. Journal of Genetic Counseling, Vol. 13, No. 1, February 2004

James Hoffman – Fragile X Ten Pin Bowler
New Zealand Special Olympics Representative

Mayor of New Plymouth, Peter Tennant, awarding James with his certificate as an Honorary Ambassador of the District.

James holding a cheque from the Fragile X Trust (NZ), which included a contribution from the NZ Organisation for Rare Disorders. Also holding a cheque from the Wellington Fragile X Support Group.

We were extremely grateful and appreciative of the kindness shown towards James' fundraising. The total amount of funds needed per athlete was $10,000 so these contributions helped in a great way towards this."

Heather Hoffman

"Come on you fellow Fragile X folks! This affliction need not be a restriction but see it as a challenge and an opportunity. We can still achieve great things." James Hoffman

Family News

Hi. Barbara Shelley here, James` mum.

How are you all? I hope you are well!

It's been an interesting time since James began his Community Participation Group called "Thumbs Up".

He doesn't want to attend every day as some days he says he has other things he needs to do, but the time he has spent there has been very rewarding, eg trips to museums, art and pottery places, and a big step for James, is actually going into a coffee bar with the group, and ordering and paying for and collecting the change for his own cup of tea! We have endeavoured to do this for many years ourselves. Trying to get him to even approach a shop counter and shop assistant has always been a huge stress to him. He is even recognising at least a $20 note, (previously he said everything was $2, even his age he said was 2!) although he has no concept of how much change or the real value of money yet, but we`re finding ways to help this as well.

Also I have been immensely helped by what I learned from the Conferences re a Fragile X person`s need for "closure", as this explains a lot of his behaviour, eg throwing out everything he thinks we have finished like drinks at McD`s as soon as we`re leaving the table whether they are finished or not, packets and things in the fridge, finished or not, stuffing washing into his drawers, clean or not, and all these other little things I never understood before. It really helps in dealing with these things, so I urge all parents to try and get along to every Conference because even though James is 23 years old I have learnt new and valuable and helpful things each time I have attended!

So a big thank you and God bless you to everyone involved in the Fragile X Trust. The work you do cannot be measured as it`s been so massive and far more beneficial than you even imagine.

God bless, Barbara Shelley

Family News

Here are a few pictures of Bradley on our recent holiday in Australia. We all had a fantastic time and have lots of wonderful memories and photos! Bradley had a lot of new experiences which he managed well (with some planning and management on the day). He even managed the corkscrew roller coaster at Sea World! Well done Bradley!"

Kim Caffell

Family News

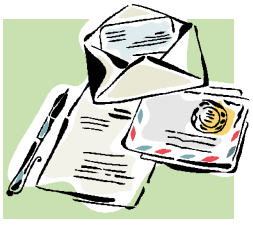
Hi everyone from a very busy Williams Family.

Life in the Williams family is about to change. Craig is actually moving out of home. He is going into a 24/7 supportive living flat with two young ladies. Craig has been with this service provider for over 3 and half years now for his work and day programme. We haven't had one behavioural problem or staff problem over this period of time. Craig always comes home with a huge smile on his face and is always laughing about something with one of the staff members as well as his mates there. The process of getting Craig into this flat has been a huge up hill battle. We first had Craig's annual needs assessment done for this last year in September 2006. The NSAC agency here at first wasn't going to play ball and I got a rather rude person saying to me that the 42hrs we get for Craig for his work and day programme is now wait for it is WHAT HE considered as a break and we could basically go get jumped on. Well my blood pressure went up and the steam started to come out of my ears. In the end we wrote to the CEO of Taikura Trust our NSAC agency here in Auckland and complained like hell. So we ended up having a meeting with e CEO and straighten things out. So now after hitting so many brick walls. We just keep chipping away and basically annoying the right people we finally got what we wanted for Craig. The ministry of Health signed off on Craig's contact and approved what we asked for. It is amazing what one gets when you don't back down and these government departments know that you are not going away and you keep annoying them no matter what in the end you there. This whole process has taken us a year and quite frankly it shouldn't happened this way. So if you hit a brick wall time and time again don't give up keep on fighting no matter what. If it is what you want and more importantly is what your son or daughter what's just dig your toes in and never ever back down. Look it doesn't matter if these government agencies get sick of your voice over phone and get sick of you writing letters just keep chipping away and you more than likely get a happy out come. Just a little tip for you all. If you run out of carers support days and you need them like yesterday. The NSAC agency you deal with your area can approve 50 carer support days over the phone without going to the ministry of health for their approval. Craig is doing very well. He is still working at the SPCA looking after the Rabbits 3 days a week and also he is back doing his gardening growing young native plants and trees with 3 of his mates at a place called Ambary Farm. The two rangers there help the guys plus a staff member from the service provider. Craig is now 26 years old and he has sure come along away since he the old days. Kevin and I are very proud of what Craig has achieved. The other night we meet up with Craig and his flatmates and the staff of the flat for dinner. We all had a wonderful time. Craig had a beer with his Dad which is neat. It is great to be able to do these things with Craig and know he is so happy and settled in his new environment. Craig still comes home every 3rd weekend as Craig puts it to see if his parents aren't misbehaving to much!!!!!. HUH as if we would !!!!.

Well the next bit of news is that we have our house on the market. After nearly 14 years of been up in the Waitakere Rangers we are heading back to the North Shore of Auckland around Torbay, Browns Bay area which we have been wanting to do for a while now. Our house is at long last all finished. So here's hoping it will not take to long to sell and to buy another house.

I do hope everyone out there is avoiding this bad flu which is going around and hopely it will not be to long before we see the sunshine again and not all this rain we have been getting.

Take care
Regards Vicki & Kevin.



FRAGILE BEGINNINGS – Pippa Wellstead's FX Story

Mark and I met in England in 1997 over the bar of Marks local pub where I was filling in time before I started my "proper job". We met, married, moved to New Zealand, started our own business, bought a house and had Jack all within 12 months of our first meeting. So, needless to say it was a very busy but exciting time for us. Jack was a gorgeous little lad. Very happy and content and so eager to please. He crawled at 13 months, walked at 17 months and loved his tucker.

When he was 20 months I read an article in a Little Treasures Magazine which talked of milestones and "what to expect". Amongst other things the article said that at 18mths a child should have a vocabulary of 20 words. Jack had only a few words, had not very good hand/eye coordination, had had numerous ear infections and he walked and ran with an unusual gait. An occupational therapist came to our home to do an assessment and her thoughts were that Jack perhaps had a trapped nerve in his hip causing him to throw a leg when he ran. She referred us to a paediatrician for an assessment.

After an hour of questioning I asked if he was going to xray Jacks hips which, to my understanding was the purpose of the visit. He folded away his notes, crossed his legs and looked me squarely in the eye and said " your son does not have a hip problem, what we have here is a boy with a severe intellectual disability who is suffering from either angelmans disease, mad puppets syndrome, autism, although this would be the least of your worries, or possibly Fragile X". He said he needed to carry out some blood tests and would forward me an appointment for the hospital. He then showed me the door and the next minute I was on The Terrace in tears with Jack crying because I was crying, wondering what on earth to do next. I had gone to the appointment alone as Mark was working and I had assumed I would be given a referral for the xrays, not to receive some devastating news like that. Thankfully a very good friend worked in Wellington and I called her in a blubbering mess and two minutes later she was there taking charge.

Naturally I went home and began surfing the net and looking into all of the possible diagnosis's whilst waiting for the blood test date to come through. Nothing arrived for 3 months during which time Jacks speech took off, he began interacting and his walking and running seemed better, so much so that I did think it had all been in my imagination. However the appointment came and we duly went to Lower Hutt hospital for a harrowing 2 hours where they pinned Jack down to extract the large quantities of blood they needed to carry out the neccessary tests. Three weeks later I received a phone call from the paediatrician saying that all the tests were clear, it had all been a storm in a teacup and to carry on as normal!

In the June - 2 months later I received a call from Lower Hutt Hospital this time from a different paediatricain, asking how everything was going and what results I had been given about Jacks blood tests. I explained that I had been given the all clear 2 months previously to which she asked if I could pay her a visit, that afternoon. I went immediately and was told that I had been given the results before the tests had been completed and that Jack did in fact have the full mutation of Fragile X.

Jack was 2½ when he was diagnosed. I was told he would be in nappies until he was 6 yrs old, he would possibly not be able to read and write, his verbal skills would be pretty limited and he wouldn't live independently. It was devastating news.

I think it is natural for any parent to want all children to have the same starting block in life and for it to be down to them as to what they make of it. Sadly this is not the case.

Jack today is 8½. He is the most gorgeous, fun loving lad with a brilliant sense of humour, who adores horses, swimming, shooting hoops, his trampoline, food, and, above all else his Grandma. He has been out of nappies since he was 3, he is reading at level 8 and can write 30 words independently. He is able to swim, he goes horse riding, goes to Keas and can imitate anyone.

Jack attends Warepa School and has just moved to the senior classroom. He has an amazing team of people working with him who have made it their aim to maximise his potential. He has a teacher aide 10 hours a week and he works to his own curriculum. His behaviour at school is impeccable and the other children are very supportive and amazingly understanding of him and his needs. At home things are a little different with us seeing the extremities of Jacks anxieties and difficulties but every day is a new one and with it comes new challenges.

We are lucky enough to have been able to extend the family and have since had two girls, Phoebe 21/2 and Emily 1. With both pregnancies I had a CVS done at 12 weeks and both were unaffected by Fragile X. Jack loves his two sisters and is extremely protective of them. There is nothing more heart warming than to hear the three of them giggling uncontrollably over some private joke.

I wouldn't swap him for the world.

The Fragile Curtain

I am acting behind frail curtains of a marquee
The crowd just watches on with compassionate glee
As I am alive in front of the flicking light
Yet the bright contact of publicity
Distracts my vision with a certain plight
To make matters worse, the script is never clear
Because when I speak the words begin to smear
Sorry, but I need to find a place to rest my head
Because all this thick concentration
Has made my skull feel like heavy liquid lead.
I sucked my hand to deflect the anxiety of the stage
Flapped my arms around like a bird lost in a cage
I've also been finding it hard to grow, to sleep
All my tinsel town expectations are a mess
All my dreams are in an itchy heap.
But in my own inner theatre
I am a playhouse king, a true trend setter
I have the best sets, the finest conductor
The greatest band on earth
I would be named a grand playwright constructor.
I could be the next Shakespeare
Gallantly being praised by my peers
With Lloyd Webber saying how my talent shined
If the outside could only step through the curtain
They too would see me raise the roof with my mind.

Daniel is from North London. He has been writing for about 14 years now on various subjects all based on experiences of life and true feelings which is what he feels real poetry is about, poetry is a form of healing. Daniel has had several online publications in the USA., Canada, New Zealand and in the UK, as well as in poetry publications. One of his poems " The Blanket" has been used in "Solutions for Adults with Asperger's Syndrome" by Juanita Lovett. He also has a website www.newdawnpoetry.freeuk.com which has been running for about four years now. He is very keen to use his poetry as a way to help families and people with disabilities.

Offering Population Genetic Carrier Screening for Fragile X Syndrome in Australia: A Research Project

Alison Archibald

When a person is diagnosed with fragile X syndrome (FXS) their family members can be offered testing to determine if they are carriers; this is called cascade testing. Cascade testing can lead to the identification of other carriers in the family and can provide those family members with information about their risk of having a child with FXS. Whilst this strategy does detect some carriers, not all carriers can be identified in this way. Transfer of information between families can be limited by factors such as distance or estrangement, resulting in some carriers not being given the opportunity to pursue testing. In addition, there are potentially many people who have FXS but have not been diagnosed, and therefore cascade testing for these families has not been an option. Therefore, many people in the general population may be unaware that they are carriers of FXS. A population-based screening program, in addition to cascade testing, will allow these carriers to be identified.

Our research involves assessing how practical it is to offer population carrier screening for FXS. A variety of studies have investigated offering population carrier screening for FXS in pregnant women but few have explored offering carrier screening to women before they become pregnant. We conducted a pilot study to assess the feasibility and acceptability of offering population carrier screening to non-pregnant women. In this study non-pregnant women who attended a family planning clinic were offered a carrier test for FXS (at no charge) and asked to complete a questionnaire on their understanding of FXS, attitudes to testing and reasons for and against choosing to be tested. Women then chose whether they wanted to have the carrier test and completed a second questionnaire a month later. A small number of women who participated in the study were then interviewed to explore their experiences.

Of the 338 women recruited into the study, 96% completed questionnaire 1 and 55% completed questionnaire 2. Of the women who took part in the study 20% chose to have carrier testing. One woman was found to be a carrier and three women had grey-zone results. Preliminary analysis of questionnaire data indicates that women's understanding of FXS was good and women were overwhelmingly in favour of population carrier screening for FXS.

A sub-group of 31 women who participated in the pilot study were interviewed to explore their attitudes in depth. This included women who chose to be tested, including those with carrier and grey zone results, and those who chose not to be tested. Test choice was influenced by: the life stage of the woman, i.e. if she was thinking about having children she was more likely to consider having the test; whether the woman had any experience with health problems, i.e. if she did she was more likely to consider having the test; and the woman's perception of the benefits of carrier testing.

Women discussed a number of themes relating to their attitudes towards screening including: acceptability of population carrier screening for FXS; importance of informed choice for pregnancy planning; impact of carrier status on life choices; attitudes towards disabilities; most appropriate settings and life stages for offering screening and implications for the wider society. These themes highlight the range of complex issues involved in population carrier screening for FXS.

Overall, our results indicate that regardless of individual choices on testing, women were supportive of population screening for FXS and felt that it should be widely available, provided it is optional and follow-up genetic counselling is available.

The next phase of our research is to conduct interviews and focus groups with other members of the community to explore their attitudes towards population carrier screening for FXS. This will include: family members of people with FXS; health professionals; pregnant and non-pregnant women and their partners; members of the school community and other stakeholders. Their responses will give us an in-depth understanding of community attitudes to population carrier screening for FXS, and will provide vital information for health planners considering the development and implementation of population screening programs for FXS.

Alison presented a poster on this research at the HGSA Conference in Auckland. She is completing her doctoral study at the Murdoch Children's Research Institute, Royal Children's Hospital, and Department of Paediatrics, University of Melbourne. Her supervisors are Sylvia Metcalfe, Samantha Wake and Alice Jaques. Jonathan Cohen and the Fragile X Alliance are also involved in the research. If you would like to participate in a similar study in New Zealand please contact the Fragile X Trust (NZ): fragilex.info@nzord.org.nz

The Marcia Braden workshops

The two workshops in Wellington and Auckland in March were a huge success. The 130 participannts were very appreciative of Marcia's depth of knowledge, her compassion and wry sense of humour. We are extremely appreciative of the effort that Marcia and husband Ken went to to fit New Zealand into their busy schedule.

The Fragile X Trust has Marcia's **Social Compass** Programme and **Maths Equivalance Boards** available for purchase. If you are interested please contact us: fragilex.info@nzord.org.nz

Disability in New Zealand – Contact Details
Needs Assessment and Service Co-ordination Service (NASC)

List of NASCs that provide services for Disability Services Directorate service users

NorthAble Northland Disabilities Resource
40 John Street, Whangarei
Ph: 09 430 0988 Fax: 09 438 9468
Noel Matthews (Manager DRC Northland)
Email: noel@drcnorthland.org.nz
Rosalie Eilering (Manager NASC)
Email: rosalie@drcnorthland.org.nz

Taikura Trust (INSA)
Level 2, 19 Charles St, Papatoetoe, Auckland
Ph: 09 277 3850 or 0800 TAIKURA (0800 824 587)
Fax: 09 278 6315 Email: info@taikura.org.nz
Sonia Hawea (General Manager)
Email: sonia.hawea@taikura.org.nz

L.I.F.E. Unlimited Charitable Trust
20 Palmerston Street, PO Box 146, Hamilton
Ph: 07 839 5506 Fax: 07 834 9982
Jane Pembroke (Manager) Email: Jane@life.nzl.org

Disability Support Link
Level 2, Monckton Bldg, Rostrevor St, Hamilton
PO Box 9201, Ph: 07 839 1441 Fax: 07 839 1225
Jan White (Manager)
Email: WhiteJV@waikatodhb.govt.nz

Support Net Kupenga Hao Ite Ora
PO Box 2121, 510 Cameron Rd, Tauranga
Ph: 07 577 3309 Fax: 07 571 0277 Cell: 021 754 474
Don Sorrenson (Regional Manager)
Email: don.sorrenson@bopdhb.govt.nz

Access Ability Taranaki
Metro Plaza, 33 Devon Street, New Plymouth
PO Box 115 Ph: 06 758 0700 Fax: 06 758 5201
Judy Bilderbeck (CEO). Tony McLean (Manager)
Email: tony.mclean@accessability.org.nz

Bay Home Support
Cnr McLeod and Omahu Rd, Hastings
PO Box 9014 Ph: 06 870 7485 Fax: 06 870 7481
Judith Peters (Manager)
Email: Judith.peters@hawkesbaydhb.govt.nz

Access Ability Wanganui
126 Guyton St, Wanganui
Ph: 06 348 8411 Fax: 06 348 0166
Wendy Kopura (Manager)
Email: wendy.kopura@accessability.org.nz

Supportlinks
69 Malden Street, PO Box 188, Palmerston North
Ph: 06 353 5899 or 0800 221 411 Fax: 06 353 5874
Maria Greig-Anderson (Manager)
Email: Maria_Greig-Anderson@enable.co.nz
DDI: 06 353 5898 or mobile 0275706152

Focus
PO Box 96, Masterton
Ph: 06 946 9813 Fax: 06 946 9826
Helene Dore (Team Leader)
Email: helene.dore@wairarapa.dhb.org.nz

L.I.F.E Unlimited
5 Bouverie St, P O Box 33-145, Petone
Ph: 04 569 3102
Marlon Hepi (Manager)
Email: marlonh@life.nzl.org

Capital Support
Level 3 Guardian House, Cobham Court, Porirua
PO Box 50-137 Ph: 04 237 2570 Fax: 04 237 2571
David Darling (Service Leader)
Email: David.Darling@ccdhb.org.nz

Support Works
14 New St, Nelson
Ph: 03 546 3984 Fax: 03 546 3983
Marie Calderbank (Manager)
Email: marie.calderbank@nmhs.govt.nz

Life Links
205 Salisbury St, P O Box 2379, Christchurch
Ph: 03 365 9593 Fax: 03 365 5244
Craig Hutchison (Managing Director)
Email: craig@casnz.co.nz
Anne Simpson (Manager)
Anne: 0274335581 Email: simpsona@lifelinks.co.nz

Access Ability Otago/Southland
Dunedin: L1, 10 George St, PO Box 966, Dunedin
Ph: 03 477 6211 Fax: 03 477 6251

Invercargill: 70 Forth St, Invercargill
Ph: 03 214 4735 Fax: 03 214 4909
Sue Hansson (Manager)
Email: suehansson@accessability.org.nz

Notice for all Parents Caring for an Ill or Disabled Child while Receiving the DPB

Are You Eligible for This Payment?

Notice from Jan Moss, Complex Carers Group

- Parents who are or have in the past been on the DPB while they are caring for a child who is sick or infirm
- Who is/was in receipt of the Handicapped/Child Disability Allowance
- Whose child would have required care elsewhere had this not been provided by the parent
- **should have been receiving the DPB Sick and Infirm benefit, which is currently $37.43 more per week than the regular DPB for sole parents.**

Three families have successfully been "back paid" substantial amounts because they were not informed that this was the case.

If you believe you may be entitled to back payment, contact the Work and Income Contact Centre 0800 559 009 and ask for the application forms for Review of Decision.
These should be sent to you to complete and return to your nearest Work and Income office.

It maybe helpful to include a letter from your GP stating that your child would need to be in a formal care facility if you were not or had not provided this care.

Please request and complete the paperwork as soon as possible. It is my understanding that once the Core Benefit commences this year, back payments will no longer be made.

If you have questions please contact me, Jan Moss, by email via info@carers.net.nz

Need to contact us?

Web: www.fragilex.org.nz

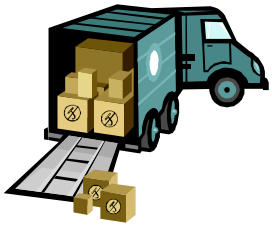
E-mail: fragilex.info@nzord.org.nz

Chairperson: Anita Nicholls
158 Oxford Terrace
Lower Hutt 5011
(04) 938-0552

Secretary: Judith Spier
196 Taita Drive
Lower Hutt 5011
judith.spier@xtra.co.nz
(04) 567-8532
021 1319778

National Co-ordinator:
Chris Hollis
158 Oxford Terrace
Lower Hutt 5011
(04) 938-0552

Change of Address?



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

Thanks!

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