What is Fragile X Syndrome?

Fragile X Syndrome is the leading inherited genetic cause of learning and intellectual disability and the most common single gene cause of autism.

Characteristics include mild to severe learning difficulties, anxiety, ADHD, sensory issues, delayed and repetitive speech and autistic like behaviours.

Strengths include strong imitative skills, good memory and visual processing, great sense of humour, empathy and a desire to be helpful.

It is estimated that as many as 1 in 468 males and 1 in 151 females are carriers of FXS.

Female carriers have a 50% chance of passing the gene mutation to each of their children. Male carriers will pass it to 100% of their daughters but none of their sons.

Some carriers maybe at risk of developing fragile X-associated disorders including late onset ataxia (FXTAS) and early menopause (FXPOI).

About 12,000 New Zealanders have Fragile X Syndrome or are carriers.
What is Fragile X?

Fragile X refers to a group of genetic conditions now referred to as “Fragile X-associated Disorders”.

The disorders include:

Fragile X Syndrome (FXS) is a genetic disorder caused by the full mutation of the FMR1 gene (a change in the DNA structure) on the X chromosome. It results in a wide range of developmental and behavioural issues and is the most common inherited cause of intellectual disability worldwide. It affects around one in 4000 individuals.

Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS) is a condition affecting some male carriers (and in rare cases, female carriers) of the premutation over age 50, causing balance, tremor and memory problems.

Fragile X-Associated Primary Ovarian Insufficiency (FXPOI) or early menopause is a condition affecting some female carriers of the premutation.

Fragile X can be passed on in families with no apparent sign of the condition. In some families, multiple generations are affected.

All children with delayed development or autistic-like features should be tested for Fragile X Syndrome.

Cover photo - Wellington Dinner with Louise Gane (far left) and Jude, Anita and Claire - May 2014
In the bad old days when my boys were young, Genetic Health Service New Zealand could argue that they were not infringing this right by refusing to test children with mild symptoms of fragile X because the diagnosis would make no difference to their care or support. However, through the efforts of our parent-led Trust, things have changed. We now offer fragile X-specific services to children, families and professionals throughout the country. We have clinics two times a year where families and their health professionals can consult with neurodevelopmental paediatrician, Dr Andrew Marshall, either in person or by video conference. Through our No Longer Fragile programme, Andrea and Anita introduce teachers and support workers to strategies that will get our fragile X children engaged and learning in the classroom. And Andrea is always on hand to provide advice and link families up with the experts here and overseas who can help address their concerns.

Now that one of our families has shown that it’s possible to make their own decision on when to test for fragile X by using an Australian lab, it’s time GHSNZ took notice and moved their service into the 21st Century. We want families to have the support of local genetic counsellors that recognise and respect the expertise and knowledge of parents. So, we have written a polite “please explain” letter to the clinical director and await to hear the outcome. If you have personal experiences of poor service from GHSNZ, now is the time you should tell us your story. Please contact Andrea or me directly.

The Hollis family look forward to seeing as many of you as possible in (hopefully) sunny Napier.

Chris Hollis
National Coordinator’s Report

With half the year already over it’s a good time to reflect on what’s been happening so far for 2014.

Of course the highlight would have to be our workshop series in both Auckland and Wellington in March with Dr Jonathan Cohen, Anita Nicholls, Rashelle Cohen, Michael Cohen and Dr Andrew Marshall. And the feedback showed that you agree. Participates seemed to particularly enjoy Michael Cohen speaking about his life and living with fragile X. It was truly inspiring to hear him speak about his achievements and the things that have helped him.

In May Louise Gane visited us and she generously spent time during her holiday with many families. I’m very happy to announce that Louise will return in November and join us for the 2014 Fragile X Family Gathering! Planning is well underway for another fun family weekend. Many families have booked accommodation already. This year we are off to Kennedy Park in Napier from 7-9th November. See below for more details.

In May we also held another Fragile X Clinical Forum with Dr Andrew Marshall. Five families around the country took part together with their local paediatrician. We aim to have another clinic later in the year so let me or your paediatrician know if you would like to take part. Remember it’s free.

Families are continuing to meet around the country and share their journey of living with fragile X. See photos later in this issue. In Whangarei Veronica has led the initiative for a group of 13 women to participate in the Auckland Marathon in November. They plan to raise awareness about fragile X and get fit in the process! If you would like to support their mission you can donate at the Givealittle event page www.givealittle.co.nz/fragilexnewzealand

We have also taken the No longer Fragile workshop to a number of schools this year and continue to receive good feedback from both families and schools.

I wonder if the second half of the year will be so action packed?

Andrea Lee

Fragile X Family Gathering

7-9th Nov. 2014 Kennedy Park, Napier

Book accommodation at www.kennedypark.co.nz or 0800 457 275

10% discount for all fragile X families

Louise Gane, from the MIND Institute will present a workshop and join us for the weekend! The program will include all our usual fun activities like the mums breakfast, the blokes night, a shared BBQ dinner, the AGM and family fun activities.
Activities and events since our last newsletter in Summer;

**Workshops**

FXNZ held workshops in Auckland on the 6th March and in Wellington on the 7th March 2014 with Dr Jonathan Cohen, Michael Cohen, Rashelle Cohen, Dr Andrew Marshall and Anita Nicholls. See pg. 7 for more information. Jonathan and Anita’s presentations are available on the FXNZ website www.fragile.org.nz

**A visit from Louise Gane**

In May Louise visited NZ on holiday to see her mother in Auckland. 13 families got together in Auckland for a fun afternoon. In Wellington 7 families went out for dinner with Louise. See pg. 13 for photos.

**‘No Longer Fragile’ Workshop**

FXNZ received funding from the Frozen Funds Charitable Trust for our education program. We have received fantastic feedback about the program and the difference it makes to school communities in understanding the strengths and challenges of people affected by fragile X.

**Facebook**

The FXNZ Facebook page www.facebook.com/Fragilexnewzealand continues to be a good way to raise awareness about fragile X and share fragile X news and events. You can visit the page without having a Facebook account. If you are a parent and would like to be part of the parents group let Andrea know.

**Auckland Marathon**

Veronica Harley has led the initiative for a group in our community to take part in the Auckland Marathon! They plan to raise awareness of fragile X and get fit at the same time. A fundraising site has been set up so if you would like to support their efforts you can at www.givealittle.co.nz/org/Fragilexnewzealand

**Genetic Health Services**

FXNZ have written to GHSNZ about issues with testing for fragile X in NZ.

**Annual Fragile X Family Gathering**

The date has been set and planning is well underway! 7-9 November 2014, Kennedy Park, Napier www.kennedypark.co.nz

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**Useful websites and links**

Useful websites for information about fragile X:

1. **Fragile X New Zealand**
   www.fragile.org.nz
2. **The National Fragile X Foundation**
   www.fragilex.org
3. **Australian Fragile X Association**
   www.fragilex.org.au
4. **The Fragile X Society**
   www.fragilex.org.uk
5. **Imagine Better**
   www.imaginebetter.co.nz
6. For information about education and behaviour www.marciarden.com
7. **Health Passport**
8. **Information about FXTAS**
   www.fxtas.org
9. **Fragile X research**
   www.fraxa.org
10. **Parent to Parent New Zealand**
    www.parent2parent.org.nz
Tyler's Song — Jenna Jones

This song is about my 11 year old brother Tyler who has Fragile X which is a type of special needs and how he's just the coolest kid in the world! Jenna Jones

Tyler's Song

He's drumming on the couch at five in the morning,
Running around with a plastic gun,
Chasing the robbers and fighting the bad guys,
Doing whatever seems fun
At the time with the perfect imagination,
Let go in the air around him,
He's got no fears,
Nothing holding him back,

His smile could light the room,
His mind could set it on fire,
This world needs more like you,
To show what real happiness is like.

I wish I could see,
The world through your eyes,
Watch the imagination run wild
Feel the flowers blooming in my heart,
And whatever name the rose will still smell as sweet
I wish I could see,
The world through your eyes,
Watch the imagination run wild
Feel the flowers blooming in my heart,
And whatever name the rose will still smell as sweet
The rose still smells sweet

Never let anyone take away your spark,
Brother; you're perfect just the way you are

From here to the sky

Jenna Jones won the Smoke Free Rockquest Lyric Award in the Auckland finals for this song!!!

Congratulations Jenna!

You can hear Jenna perform her song on YouTube and follow Jenna Jones musician on Facebook
Around 100 people attended our Fragile X Workshop series in Auckland and Wellington. The presentations are available to download on our website www.fragilex.org.nz for those who couldn't make it. Below is a brief summary of some of the information presented by Dr Jonathan Cohen about Fragile X Syndrome.

**Interventions**
- Multidisciplinary team;
  - Audiologist
  - Optometrist / ophthalmologist
  - Speech and language therapist
  - Occupational therapy / physiotherapy
  - Psychologist / psychiatrist
  - Specialist – paediatrician, neurologist, clinical geneticist, genetic counsellor
  - Teacher / Special education
  - General practitioner

**Checklist**
- DNA testing to confirm FXS status
- Genetic counselling for information and cascade testing of relevant family members
- Grief and supportive counselling for family
- Hearing assessment with audiologist
- Vision assessment with optometrist / ophthalmologist
- Assessment for orthotics with podiatrist
- Speech & language therapist for help with communication
- Educational psychologist - assess ADHD and ASD, behaviour management strategies
- Occupational therapist including sensory issues
- Developmental paediatrician for initial assessment and ongoing review
- Trial medications for anxiety or ADHD if present under care of medical practitioner
- NASC referral for help with funding, home support etc. & Child Disability Allowance if appropriate
- Give information on FXS and advise to join Fragile X New Zealand (FXNZ)

**Multidisciplinary assessment at FXS Clinic**

**Treatment**
- Medical - epilepsy, CT problems
- Pharmacological - ADHD, anxiety, mood, urinary, sleep, aggression
- Behavioural - ADHD, anxiety, depression
- Educational - strengths and weaknesses

*Specific treatment and management strategies are of great benefit to individuals and their families*

**10 top things for Teachers to know**
1. Don't force eye contact
2. Expect inconsistency
3. “Simultaneous” vs “sequential” learners
4. Allow and/or encourage frequent breaks
5. Verbal expression is cognitively taxing. Think “indirect”
6. Prepare for transitions
7. Work with an OT to embed Sensory Integration into the day
8. Notice environmental triggers
9. Know FXS strengths
10. NB. Students prone to hyperarousal & anxiety

Also see Lesson Planning Guide
(Laurie Yankowitz, www.fragilex.org)

Rashelle Cohen presented information about vision issues and fragile X. She stressed the importance of people with fragile X having regular thorough eye and vision checks with a behavioural optometrist.

FXNZ has a list of recommended behavioural optometrists if you are unsure who to see in your area.
Trialling an SSRI for Anxiety

Late last year when our son Connor was 7 years and 4 months we went to our paediatrician to talk about trialling him on an SSRI (selective serotonin reuptake inhibitor) to help with his anxiety. After talking to different people in our fragile X community who were getting good successes using an SSRI, we wanted to trial Connor.

After some investigation and consultation by our paediatrician it was decided to trial fluoxetine. Very quickly Connor became highly activated, wasn’t sleeping and was generally bouncing off the walls! Connor is a very active child without adding to it, but he had normally slept well, so you can imagine we were not happy with this change!

We did however also see some good things we hadn’t seen before while trialling the SSRI. For example he got up on stage and did some highland dancing at the Highland Games at school and generally managed tasks better in class and at home. He could manage to finish a task with two instructions.

However the activation and increased hyperactivity was too much. We went back to our paediatrician with some addition information from FXNZ and she agreed to trial Sertraline (Zoloft). We started on a very low dose - 1/8 of a 50mg pill. We quickly saw great results but with much less activation and hyperactivity!

We feel Sertraline is helping Connor be the best that he can be. It helps him be open for learning. The school now say they can get up to one hour at a time of learning. This is a sample of his handwriting after taking an SSRI. Before we just got his name!

Sertraline is only one of the tools and strategies we use to help Connor. But it helps the other strategies to become more useful and effective.

In hindsight I would have pushed harder to trial Sertraline first. Dr Randi Hagerman suggests that it often works well for treating anxiety in fragile X. It was a hard choice to make to start Connor on an SSRI but with information and talking to other parents in our community Chris and I are happy with our decision and happy with the results for Connor. Thank you Andrea and our lovely fragile X families for your support and help!

Vicki - Whangarei
Connor competing in the Raurimu Avenue School Triathlon!!!

Congratulations from FXNZ Connor!

Fragile X Clinical Forum

The Fragile X Clinical Forum is an opportunity for a discussion via video conference link between families affected by Fragile X Syndrome, paediatricians responsible for the local care of children with Fragile X and Dr Andrew Marshall, Developmental Paediatrician and specialist in Fragile X Syndrome.

This is a free service available to all families. Families can use the service more than once as the need arises.

Please contact FXNZ for more information or to register for the next clinic.
Working with your NASC

Understanding how the NASC service works and how to work with the service will ensure you achieve the best results for your child and your family.

Chances are if your child has a diagnosis of fragile X your family will have come across the Needs Assessment and Service Coordination (NASC) service. There are 15 NASC’s across the country. In each region they are known by a different name, e.g. in Auckland the NASC is called Taikura Trust.

The NASC’s role is significant because they control all Ministry of Health (MoH) funded support for people with disabilities. The NASC doesn’t deliver ‘hands on support’. Their role is to determine who is eligible to get services and the type and amount of service the person gets. Essentially this is a gate-keeping role.

The MoH eligibility criteria for disability services is: “A person with a disability is a person who has been identified as having a physical, psychiatric, intellectual, sensory, or age-related disability (or a combination of these), which is likely to continue for a minimum of six months and result in the reduction of independent function to the extent that ongoing support is required” (MoH). The good news, our fragile X children with an intellectual disability clearly meet criteria and some of the girls without an intellectual disability may meet criteria for a sensory or age-related disability.

Many of you will have gone through a Needs Assessment with your NASC. Usually this is a meeting at your house to gather information about your child and the support needs they have. We recommend you have another person there with you during this meeting as the support package - the amount and type of support you receive - is all dependent on what needs are identified at this meeting. You need to be very clear about the times your family or child needs extra support. For example we know that children and adults with fragile X often struggle to establish and maintain friendships and participate in everyday events with their peers. It’s appropriate to stress this fact so that the support package gives your child opportunities to get out and have fun doing what they enjoy doing (even better if the activity they are supported to attend is with their peers).

Make sure you use the support to get a real break for yourself. For example Anita and Chris in Wellington plan to use their Carer Relief Days to pay for someone to look after their two FX boys, Ben and James, in their own house over a weekend. Anita said “I realize now that Mum’s gone we have no one that could just step in an emergency to look after the boys. I feel safer knowing that there is someone down the road who knows their routines well enough to look after the boys overnight in an emergency.”
Based on the Needs Assessment your child will be allocated a support package. For a younger child this might include a Carer Support allocation in the form of days per year. As children get older (around 7 years up) this might also include a Personal Support package, or potentially Household Management) allocated in hours per week, a residential respite service or a holiday program. Alternatively you may choose to use your allocation through Individualised Funding (IF) via a fund holder such as Manawanui InCharge. This means you choose who you employ to provide the services you need. You may also be able to request support from a Behavioural Specialist service.

The types of support you can ask for vary around the country (it shouldn’t but it does) so it’s really important to understand what is available in your area so you know what you can ask for.

Tony McLean gives this summary of working with the NASC and navigating the system, “In theory, each person should be able to access supports and services to meet their individual disability-related needs regardless of where they live. In reality, however, there are regional differences in funding allocation, services, service quality, and information that make navigating the system complex, challenging, and confusing for many people.”

If you have found this process confusing and complicated you are not alone! We suggest getting as much help and guidance as possible to get the best result. Our national coordinator Andrea Lee is available to support you through this process or connect you with a family close by to find out more about services in your area. We also recommend Imagine Better’s Pre-Assessment Plan service. Find more at www.imaginebetter.co.nz or contact them on 0800 787 587.

**Why do VISUALS help?**

Visually-based SUPPORTS work for a number of reasons:

People with FXS are visual learners. Take advantage of this strength!

Anxiety is a significant problem. Verbally explaining the days' routine, or verbally describing what will happen at the dentist's office may work to increase anxiety, as working memory deficits make it difficult to hold information in mind. Presenting information visually - for example, through use of a visual schedule, can alleviate this problem.

Visual Supports help “translate” the environment and expectations to a person with FXS without requiring direct processing of language. This type of information can be processed quickly. Visual supports remain present, so they can be referred to more than one time, whereas verbal directions might be heard and then forgotten.
Good news first, our daughter Charlotte is attending Hauraki Plains College, as any other normal student.

When we got her diagnosis she was 9 years old, and we knew her social skills were on the whacko end of the scale, we were (well, I was) petrified of her navigating the teen years in the rough oceans of high school.

She is now in one good academic class and getting good grades. One of her subjects is French (I’m French, she’s bilingual) so that’s super easy for her (take the grades where you find them!). Anyway she is better with her brain than she is at handiwork, so it’s just as well she has now decided she wants to become a lawyer or something like that (a change from Queen, or Circus Artist), and she may well make it happen, who knows...

Give the parents some credit, we did pick the strictest school in the area, with old fashion values of good behaviour, expectations of hard work, rules and boundaries announced clearly and enforced seriously. But also a commitment to the students, to getting to know them well, and helping them shine in their particular abilities. This was to reassure us (well, me) but it also was a choice to make life easier for Charlotte, who likes it clear and consistent and thrives on structure.

Charlotte so far has managed to keep her own fragileXness hidden from her fellow students. Some of them must realise that she is a bit different, of course, but she has made some good friends. And I like the ones she’s picked. Maybe they all are a bit strange at that age!

She does not want to have to explain and justify fragile X about her self, but she does not mind talking about her brother.

She agreed for us to share it with the adults at the school, which we would have done anyway. We thought that she may need a bit of special attention and care with her anxiety and concentration, as well as the fact that she does not ask for help when she gets stuck, because she is... stuck. Well, the dean made me write a little blurb on Charlotte’s particular learning style and idiosyncrasies, as well as what the school could do to help and accommodate her. Then she passed it to all her teachers. Every time we come in contact with one of them they seem to have read it and they talk about our girl like they really know her and dig her. Oh yeah!

We never thought she was going to pick up any sport. She looked for many years like one of those kids not even interested in the internet, as she was already in her virtual world of faery tales and imagination, since she had discovered BOOKS when she was 2, and never looked back. Once again she surprised us all, she has become a rower, of all things. And she has discovered the internet and social media (but that’s another story!)

Apart from that she is also a helpful teenager with minimal attitude and mood swings (thanks to added molecules), looks after her brother, and my rabbits, and does chores in the house, and is good company to go out on a coffee date with, and has the same size shoes and clothes as I do (for now).

So that’s Charlotte, looking on the sunny side.

Anne - Thames
Community News

Above; James, Phil, Liam, Grayson, Ben and Danielle at dinner with Louise Gane in Wellington

Right; Bogdan, Mirela, her mother and Louise Gane at the family gathering in Auckland

Right; Claire and Vinnie at a family get together at Paekakariki Beach!

Left; Antionette, Patricia and Leona at the Auckland family gathering with Louise Gane

Right; Dan Coe at the International Fragile X Conference in the US with Jonathan, Michael and Rashelle Cohen, our Australian FX friends!
FXPOI - Fragile X-associated Primary Ovarian Insufficiency
FXPOI is one of three known Fragile X-associated Disorders (FXD). FXPOI is a condition in which the ovaries are not functioning at full capacity in an FMR1 premutation carrier. Common symptoms of FXPOI include absent or irregular periods, symptoms of menopause such as hot flashes, etc., early menopause and infertility. FXPOI occurs in about 20-25 percent of adult female FMR1 premutation carriers.

How does FXPOI differ from Early Menopause?
Even though women with FXPOI may develop symptoms similar to those of menopause, such as hot flashes and vaginal dryness, FXPOI differs from menopause in some important ways:
Women with FXPOI can still get pregnant in some cases because their ovaries may occasionally function to release viable eggs. Women who have completed menopause cannot get pregnant because their ovaries no longer release eggs.
Women with FXPOI can experience a return of menstrual periods. Women who have completed menopause will not have menstrual periods again.
FMR1 premutation carriers can have normal ovarian function, but can still go through early menopause, which is menopause occurring between 40 and 45 years of age (menopause normally occurs between 45-55).

What is POF and how does it differ from FXPOI?
POF stands for “premature ovarian failure,” which is the complete cessation of menstrual periods before the age of 40. The range of FXPOI symptoms includes POF, which is at the more severe end of the spectrum. FXPOI also includes other symptoms of ovarian insufficiency including irregular cycles, “sub-fertility” or infertility, hot flashes, etc. All of these encompass a wider range of symptoms than POF alone.

How do you know if you have FXPOI?
If you are a FMR1 premutation carrier, you should alert your doctor to the risk of FXPOI in carriers (some physicians might not be aware of this condition). All female carriers should keep a log of their menstrual cycles and note any unusual patterns, missed cycles, abnormally long cycles (more than 35 days) or short cycles (less than 21 days). Also note any other physical changes such as hot flashes, insomnia, vaginal dryness and/or decreased libido, increased anxiety, etc. If you are concerned about FXPOI, talk to your doctor about having your FSH, a hormone that reflects ovarian function, measured. FSH levels increase as you approach menopause or if your ovaries are not functioning properly for your age.

With thanks to;

www.fragilex.org
Fragile X New Zealand have a lending library that includes books, audio-visual material and educational information about fragile X.

We regularly update our stock with newly published books. We try to profile these new books in the newsletters and you can view the full list of books, and other material available on our website.

www.fragilex.org.nz

Tracey Cockcroft kindly operates the library service. The loan conditions are as follows;

1. There is no limit to the number of items that can be borrowed at one time.
2. We cover the costs of delivery to the borrower.
3. The borrower is required to pay the costs of returning borrowed materials to the library.
4. Loans are for one month unless special arrangements are made.

Because this is a free service, we request that all care is taken to ensure that loaned material is returned in good condition.

Fragile X New Zealand does not necessarily endorse the contents of all the publications in our library.

This is an absolute must read for anyone who lives with, is related to, teaches, cares for or is interested in fragile X!

Cindi Rogers is a rock star of the fragile X world and has been an inspiration to families around the world for many years now.

What I like about this book is it’s easy to read, very engaging and heart warming, inspiring and educational all at the same time.

Through sharing the story of their families journey (their son’s are 23 and 26 now) we learn a lot about the way fragile X affects people and the strategies, interventions and adaptations that make a real difference in the lives of people affected by fragile X.

Most of all her family’s story offers hope for a happy and good life!

Andrea Lee  A copy of this book is available in the FXNZ Library
If you are moving house, changing phone or email address please contact us!

Do you have something of interest to put in this Newsletter?

Please send your articles to nationalcoordinator@fragilex.org.nz

Remember, this is your Newsletter. We want to hear about what's happening in your household, at your child's school or in your local community, so that we can share it with our national community. **Deadline for the next issue: Monday 15th Sept. 2014**

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**Donations help us do our work supporting families living with fragile X and associated disorders and raise awareness about the disorder.**

**Donations can be made to the FXNZ account ASB 123152 0096420 00**

Donations over $5 are tax deductible, receipts available

**FXNZ Wish List….**

Funding for copies of 'Understanding Fragile X Syndrome' books for newly diagnosed families

Funding to support families to attend the Family Gatherings for the first time

Funding to support the No Longer Fragile Workshop in schools

Website developer to redesign FXNZ website. Email Andrea at info@fragilex.org.nz

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**Sincere thanks to our generous sponsors -**

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