



# xpressions

## Fragile X Research Foundation of Canada Newsletter

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## What is a Clinical Trial?

By Carlo Paribello M.S.M., M.D., President, Medical Director, FXRFC

“The goal is to identify “therapeutic targets” so that new chemical substances or treatments that act on these targets can be synthesized and evaluated.”

Over the past several years, we have reported several significant breakthroughs in Fragile X research and used terms like “therapeutic targets” and “clinical trials”. However, as a father of two sons with Fragile X, I can understand a parent’s frustration with the seemingly slow pace at which these laboratory breakthroughs are translated into something we can actually use to treat our children. The development of any new drug or treatment starts with years of basic research by scientists in order to gain an understanding of the molecular

and cellular basis of the disorder being studied. The goal is to identify “therapeutic targets” so that new chemical substances or treatments that act on these targets can be synthesized and evaluated.

The next step in evaluating potential new treatments is to test them on animals. These are also called “preclinical trials”. Animal testing is an important and necessary step in developing any new treatment. Such tests show whether a potential drug or therapy has toxic side effects and what its safety is at different doses. The results then point the way for human testing.

Most of us understand that drugs or other forms of therapy intended to

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## What is a Clinical Trial con't.

treat people have to be tested on people. These tests, called “clinical trials”, determine if a drug is safe and effective, at what dose it works best, and what side effects it causes. This is information that guides health professionals and, for nonprescription drugs, consumers, in the proper use of medicines.

Clinical trials of experimental drugs proceed through four phases:

- In Phase I clinical trials, researchers test a new drug or treatment in a small group of “normal” volunteers (20-80) for the first time to evaluate its safety, determine a safe dosage range, and identify side effects. This usually takes about 2 years.
- In Phase II clinical trials, the study drug or treatment is given to a larger

group of people (100-300) that have the medical condition that you want to treat, to see if it is effective and to further evaluate its safety.

This also takes about 2 years.

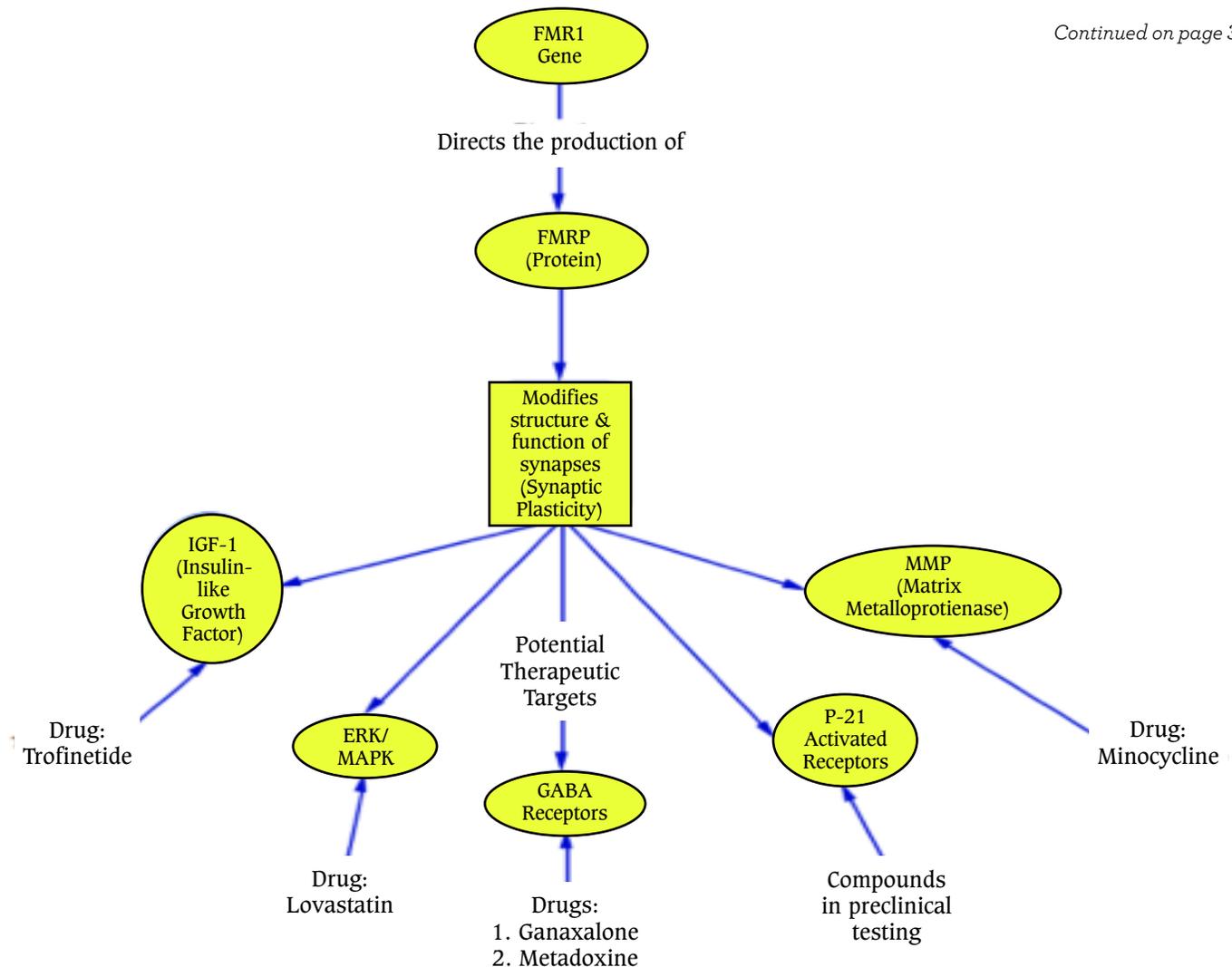
- In Phase III studies, the study drug or treatment is given to large groups of people (1,000 - 3,000) to confirm its effectiveness, monitor side effects, compare it to commonly used treatments, and collect information that will allow the drug or treatment to be used safely. This may take 3 ½ to 5 years.
- Phase IV studies are done after the drug or treatment has been marketed and take an additional 2 years. These studies continue testing the study drug or treatment to collect information about their effect in various

populations and any side effects associated with long-term use.

Controlled clinical trials, are a type of trial in which results observed in patients getting the drug are compared to the results in similar patients receiving a different treatment. They are the best way science has come up with to determine what a new drug really does. That's why controlled clinical trials are the only legal basis for Health Canada (and the FDA in the U.S.) to conclude that a new drug has shown “substantial evidence of effectiveness.”

Fortunately, the years of basic research into Fragile X have begun to pay off! Over the past several years, a number of potential therapeutic targets have been identified (Figure 1 below).

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Since the FMR1 gene was first identified in 1991, efforts to treat Fragile X have aimed at three different levels:

- The first is to find ways to turn the gene back on (a cure),
- The second is to try to get FMRP into brain cells,
- The third is to synthesize drugs that can compensate for the loss of the FMRP Protein (a treatment).

Most of the progress that has been made over the past 5 years has occurred in the latter of these two approaches.

However, since the farther “downstream” we get from the gene, the more the pathway diverges. This in turn means that a greater number of “therapeutic targets” will have to be treated in order to compensate for the loss of the FMRP Protein. Therefore, the first effective treatment will likely involve a combination of several new medications which target specific neuronal receptors and signalling pathways within the brain cells.

Recent discoveries made by Fragile X syndrome researchers are some of the most important scientific discoveries of

the last 20 years! Fragile X researchers are now testing compounds which have a specific effect on the core deficits of Fragile X syndrome. Some of these compounds have now made it into the drug “pipeline” and will be developed to treat Fragile X. Below is a list of ongoing or recently completed clinical drug trials, and a complete list can be found at [www.clinicaltrials.gov](http://www.clinicaltrials.gov). We think that it is a realistic near term possibility to have routine treatment for Fragile X, that is, not just the symptoms of Fragile X, but the core features of Fragile X, within the next few years!

### Trials and Studies Awaiting Results

- February/2016 - Double Blind Crossover Trial of Ganaxolone - sponsored by our U.S. counterpart, the FRAXA Research Foundation.  
*Investigators: Frank Kocoy and Anke Van Dijk- Antwerp, Belgium*  
This trial has completed recruitment and results are being analyzed
- February/2016 - Clinical trial of Ganaxolone in children with Fragile X - sponsored by Marinus Pharmaceuticals at the MIND Institute, UC Davis. This trial is still running but recruitment is complete
- 2012-15: Epigallocatechin gallate (mega green tea extract) – *Mara Dierssen - Barcelona, Spain.*  
Trial results are being analyzed

### Completed Clinical Trials With Results

- December/2015 - Neuren’s Trofinetide showed some success in small Phase 2 clinical trials in Fragile X syndrome and in Rett Syndrome.

## Mark your calendars - July 20-24, 2016 The 15th International Fragile X Conference in San Antonio, Texas at the Marriott Rivercenter!



The National Fragile X Foundation’s (United States) biennial international conference presents a unique opportunity where researchers and parents can interact throughout the conference. Many researchers have commented on the experience of socializing with the parents of children with Fragile X, and how the experience has motivated them or reinvigorated their enthusiasm to make a difference in the lives of persons with this disability. The majority of the conference sessions are “Family-Friendly” and cover all three Fragile X-associated Disorders (FXS, FXTAS, FXPOI), with the greatest number of sessions focusing on FXS. In addition to the family-friendly sessions there are many scientific and technical sessions. Some of the benefits from attending include:

- Discover the latest research by some of the world’s leading Fragile X experts.
- Learn new techniques, behavioral interventions and coping strategies.
- Connect with researchers, clinicians, educators, psychologists and parents.
- Get the support you need.
- Start new lifelong friendship and reconnect with old friends.

You can register online by going to: [www.fragilex.org](http://www.fragilex.org) then clicking on “International Conference” and follow their directions to the registration page.

# It's the 13th Annual FX Foundation Family Fun Day!!



**Sunday June 19th 2016, 11am-2pm**

Relax and enjoy a wonderful day of fun & fantastic gourmet food (we mean free hotdogs, burgers, veggie burgers & drinks) along with other incredible FX families! Feel free to bring along aunts, uncles, grandparents or whoever supports you & your family. This is your chance to meet other families, and let the kids play at a great facility. There is a fenced-in outdoor green space & patio as well as indoor space in case the weather does not co-operate.

Please RSVP no later than **June 12th**. Contact Lori Beesley (info. next page) and state how many adults & children are coming in your group.

Bring along your smiles and folding chairs if you have them (but not your pets) and get ready to have a great time, with lots of fun and laughter! There will be lots of play equipment provided, a 50/50 draw, a bubble blowing station & more!

*This year we are again holding a lucky draw fundraiser with great prizes, so bring your wallet! If you have connections & could donate a gift card, tickets to a show or event, or a fabulous prize, please contact Lori asap or by June 10th.*

*We are asking each family to contribute one dozen peanut-free treats for the dessert table if you are able - YUM!*





**Variety Village is located at 3701 Danforth Avenue Toronto, Ontario M1N 2G2**

for more information on Variety Village, visit [www.varietyontario.com/village/](http://www.varietyontario.com/village/)

Please note that you can ONLY exit south off the 401 at Warden Ave. or Brimley Rd.

Also be aware that Danforth Rd. & Danforth Ave. are both in this area.

Variety Village is on Danforth Ave. It's on the south side, and sits next to a high school.

You can park in the school lot if the Village lot is full, then follow the walkway to the Village.

There is a set of traffic lights at Variety Village, so just turn in where you see the sign out front!

**Remember to let Lori know by June 12th!**

**Contact Lori Beesley at 416-261-2666 or email: [fxpicnic@fragilexcanada.ca](mailto:fxpicnic@fragilexcanada.ca)**

Please specify how many adults & how many children when you call or email.

Please remember to leave a number where you can be reached, just in case.

Due to limited seating, please bring folding chairs if possible.

**We are looking for teenagers to volunteer the day of, so please contact Lori early if you know someone that can assist. They can earn time toward their high school community hours!**

**Thank you and hope to see you on June 19th!**

## 3 Athletes - 2016 Canadian Special Olympics, Winter Games, March 1-5 in Corner Brook NL

**David Jessome**, proudly wears the Bronze Medal, which he earned in the Giant Slalom - intermediate level at the 2016 Canadian Special Olympics, Winter Games held in Corner Brook, NL this year. Mom, dad, and sister Catherine, watched proudly as David skied his way to the podium.

An impressive opening ceremony marked the official start of the games, and at the end of the games, there was a closing ceremony. Meanwhile, over the course of 5 days, 650 athletes from all the provinces and the territories competed in the following 7 sports: curling, floor hockey, figure skating, speed skating, snowshoeing, alpine skiing and cross-country skiing.

Despite the varied weather conditions, heavy rain squeezed between two snow storms, the mission staff, as well as several coaches and volunteers were dutifully on site to supervise the athletes and to ensure the smooth running of each event.

All in all it was a great scene, the athletes cheering on their fellow team members, meeting new friends. Parents mingled with other parents as well as being treated to a wonderful evening of local folk songs and some good down home cooking. Just a fantastic time was had by all...

*David proudly shows his well earned medal.  
Congratulations David!*



**Benjamin Paribello** and his brother **Nicholas Paribello** recently attended the Special Olympics Canada Winter Games in Corner Brook, Newfoundland, representing Team Ontario in Nordic skiing. The Blow Me Down Cross Country Ski Trails were the perfect venue, although the snow didn't always cooperate. At one point, 70 local people came in to shovel snow onto the trails!

Battling the frigid temperatures, Benjamin won a bronze medal in the 500 metres classic. Older brother Nicholas, competing in the top division, placed fifth in the 5km skate skiing event, breaking the 30 minute mark for the first time and finishing in 28:53, a personal best!

The athletes now anxiously await the roster for the Special Olympics World Winter Games to be held next year in Graz, Austria.



*Congratulations to Benjamin (left) and Nicholas (right) on their outstanding achievements in Corner Brook!*

# The Victoria Fragile X Conference

## University of Victoria, B.C. in November 7, 2015

*By Tessa and Lawrence Spero. Lawrence is a Board Member of the FXRFC*

A very focused group of 85 people, including families and their support circles, educators and health care professionals attended the Victoria Fragile X and Related Conditions Conference. Speakers included Dr. Carlo Paribello of the Fragile X Research Foundation of Canada; Tracy Muran-Stackhouse, Occupational Therapist and Sarah Scharfenaker (known to many as “Mouse”) Speech and Language Pathologist, both from Denver, Colorado; Dr. Elena Lopez, Sunny Hill Health Centre for Children, Vancouver; Gudrun Aubertin, Victoria, Clinical Team, and Kirsten Madsen, FXS Parental Liason, Nanaimo.

### *Tessa writes: What I learned at the Conference...*

- The main thing I learned was that there are an awful lot more people with Fragile X than I had thought. There are thousands. I was very sorry to learn that. We live on an island with a population of less than 400 kids, and there is 1 other school aged person with Fragile X that I know of, and, I think there may be 1 more.
- I learned that how my husband and I had described our son the whole time was valid. We had been to millions of conferences about Fragile X in the past and they did not have anything new that we could bring back to his teachers. So we stopped going. Now he is 34 and they have Relevant Jargon! So his 3 year old meds regime tackles “Hyper-arousal”. That explains why he so often flies off the handle with no warning and how he sleeps like a dead dog when he does sleep - he has an Official Description.
- We all participated in a demonstration of hyperarousal. We formed groups of 3, and 2 of us yelled and demanded that the 3rd person do things. It was awful for all of us, no matter which position we had. It was just chaos and everyone wanted to run away.
- There was a great presentation on learning style! For example, to make a snowman with various craft baubles, many teachers show the final result and then give the kids the first items to glue. Then the next and by the end they have a screaming kid and they can't figure out why. The presenter suggested showing the final result and putting out all the items on the table and letting the kid go to it. The kid can do it in any order he likes and it will be done. Now that particular example would probably work with everyone, but if you expand that idea out to other examples, it's interesting. I think people are so careful with Special Needs kids.
- People with FXS are "Incidental Learners". That is how our son learned lots in his segregated classrooms. Everyone else would be around a table and he would often be in the kitchen (same large room, different area). The teachers would always laugh when he supplied the correct answers when no one thought he was paying attention, so they let him stay in the kitchen. Maybe jargon can give the teachers credibility when they let kids do other things.

I enjoyed this day-long conference immensely. I liked meeting people in the same boat as I am in. There were a lot of O.T.s, some teachers, and not enough parents as usual. The organizers had enlisted special ed students to provide day care. Lots of attendees asked that next year this conference move around the area to make it easier for people who live further away to attend.

### *Lawrence Spero writes: What I learned at the conference from Tracy and Mouse*

- People who have Fragile X learn a whole process simultaneously not sequentially. They want to see the whole video rather than a sequence of flash cards. Show end product - gestalt learning.
- Math and numbers are hard but if you replace numbers with images - three cats, four dogs etc. it works much better.
- Use indirection for teaching. Talk to the child sitting next to them rather than directly at them. Try to use “happy talk”, be positive not negative. Use Incidental learning - side dialogues. Take emphasis off the individual put it on the visuals.
- Use beginnings, ends and timetables and “how do I know when I am done” then “what should I do”.
- Build on their huge imitations skills.
- People with Fragile X live in a state that we would describe as hyperaroused. Normal for them would be hyperaroused for us. Work on *Ready - Not Ready* regulation. Increased perseveration is a good indicator of increased hyperarousal. Approach/avoidance - walking towards at the same time as pulling back also reflects hyperarousal.
- Bring what we know to adult services! This was something they said all parents should do with out further explanation. It was one of the few times a professional implied that parents know more about their kids than they do.

I look forward to next years' conference.

## Important Information

# Sign up now to get this newsletter by email! This is the 2nd last print issue!

Canada Post increased first-class postage prices significantly in March 2014. This has had a financial impact on the FXRFC because it increased our cost to mail out both our newsletters and tax receipts. Due to this increase and other costs involved in delivering this newsletter in a printed format to families across Canada, we have made the decision to move to an **E-Newsletter format**. There will be only one more printed newsletter after this issue in order to give everyone time to sign up and send us their e-mail address.

**Don't wait! Send your E-mail address please!**

You can sign up to receive this newsletter via your email in-box NOW!

We ask you to share an email address with us, and you will receive a colour newsletter in an easy print-at-home pdf format! Or save a tree and read on-line! Just email [info@fragilexcanada.ca](mailto:info@fragilexcanada.ca) with your preferred email address and your next FXRFC newsletter will arrive via your in-box.

*Thank you for understanding!*



# FRAGILE

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## Mark your calendar

### FXRFC Family Fun Day

**Sunday June 19th in Scarborough, ON**

Join us for this annual event. See pages 4 & 5 of this newsletter for all the details and come have some fun! Please email:

[fxpicnic@fragilexcanada.ca](mailto:fxpicnic@fragilexcanada.ca)

by June 12th to let us know if you can attend and how many people you are bringing. We are also looking for teenage volunteers (earn Community Service hrs.) so contact the email above if you know anyone who can help!

### Scotiabank Runs for FXRFC

**Sunday June 26th in Vancouver, B.C.**

**Sunday October 16th in Toronto, ON**

**Come & participate!**

Fragile X Team needs you! Run or walk! It is a lot of fun for the whole family. If you can't run, join the cheer squad and yell for the team! To join the BC Team contact: Carrie Lakatos at [carriework@shaw.ca](mailto:carriework@shaw.ca) To join the Toronto Team, contact: Ian Shearer or Jennifer Williams at: [fundraising@fragilexcanada.ca](mailto:fundraising@fragilexcanada.ca) We'd really like to get teams in Calgary and Montreal! Anyone who lives in those cities and could lead a team, please contact us and we will help & support you!

If you are not able to join us, please consider helping by sponsoring a FX Team member. Close to the dates, you can go to the Scotiabank site link & sponsor a FXRFC Team. **Go FXRFC Teams Go!!**

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