



xpressions

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International Fragile X Researchers Meeting in Quebec

From October 12-18, 2017, researchers from Canada, the U.S., Europe, and Australia converged at the Hôtel Sacacomie in Quebec for the 18th International Fragile X and Related Neurodevelopmental Disorders Workshop.

This workshop was partly sponsored by the Fragile X Research Foundation of Canada and FRAXA, and emphasized recent breakthroughs in our understanding of intellectual disability (ID), autism spectrum disorders (ASD), and related neurodevelopmental disorders such as Fragile X syndrome. These biennial meetings have already lead to the discovery of many disease causing genes, and the development of novel therapeutic strategies for these devastating disorders.

After the disappointing setbacks with the Novartis and Roche Clinical drug trials for Fragile X in 2014, researchers have regrouped to try to uncover new approaches to treat this disorder. The emphasis has now shifted to “drug repurposing”. That is, finding existing drugs whose mechanism action just happens to overlap with the abnormal processes that occur in the brain cells of people with Fragile X. In addition to minocycline (an antibiotic) researchers have identified several existing drugs that may improve the symptoms of Fragile X.

These include two diabetic drugs (Metformin and Pioglitazone), an anti-inflammatory (Sulindac) and a cholesterol lowering drug called Lovastatin.

Since each of these drugs affects different chemical pathways in each cell, it is now believed that various combinations of these drugs would be the most beneficial treatment and that the best combination may vary from patient to patient. Dr. Francois Corbin from the University of Sherbrooke, Quebec reported on his “LovaMix” clinical drug trial in which he treated 20 people with Fragile X (13-40 years old) using Lovastatin (a cholesterol lowering drug) and minocycline (an antibiotic). He reported that there were significant improvements in anxiety, behaviour and social responsiveness during treatment. Work is still ongoing to determine if the effect of these drugs on brain activity and neurochemistry can be measured using functional magnetic resonance imaging of the brain (sMRI/fMRI) and transcranial magnetic stimulation (TMS), to assess synaptic plasticity (LTP, LTD) in patients.

“All children demonstrated improvement in language and or behaviour as measured by a parent questionnaire before and after treatment.”

EARLY BIRD DEADLINE EXTENDED TO SEPTEMBER 1/17!
18th INTERNATIONAL
Fragile X and Related Neurodevelopmental Disorders
WORKSHOP

October 12-16, 2017
Hôtel Sacacomie, Canada



The 18th International Fragile X and Related Neurodevelopmental Disorders Workshop will be held at the Hôtel Sacacomie, in the beautiful province of Quebec, Canada.

The Workshop will focus on delineating the genetic causes and underlying gene mechanisms associated with inherited intellectual disability.

Dr. Sébastien Jacquemont
CHU Sainte Justine
University of Montréal

Dr. Jacques Michaud
CHU Sainte Justine
University of Montréal

Dr. David Picketts
Ottawa Hospital Research Institute
University of Ottawa

Email for more information: XLIDWorkshop@ohri.ca
www.18XLIDWorkshop.com

Dr. Nahum Sonenberg from McGill University reported that Metformin (a diabetic drug) reversed the core deficits seen in the Fragile X fruit fly model. Based on these preclinical findings, Dr. Randi Hagerman conducted a small pilot drug trial using metformin on six children aged 2 to 6 years old. All children demonstrated improvement in language and or behaviour as measured by a parent questionnaire before and after treatment.

Dr. David Hampson at the University of Toronto, an FXRFC funded researcher, reported his promising finding using gene therapy to correct the brain abnormalities in Fragile X mice. His work involves the packaging of a normal, functioning copy of the FMR1 gene (transgene) into a specialized virus (AAV, or Adeno Associated Virus). Dr. Hampson's team then injected this virus into the cerebral spinal fluid surrounding the spine of the Fragile X mouse, using a technique that is similar to the spinal anesthesia that some women have done when they deliver a baby. The AAV particles then circulated around the entire brain of the mouse, allowing it to transfer its normal FMR1 gene into individual brain cells. This resulted in a correction of the abnormal behaviours and protein production in the Fragile X mouse. Dr. Hampson's current work is directed towards optimizing the amount of FMRP produced by the transgene, in order to mimic the naturally occurring production, in the Fragile X mouse. The ultimate goal is to create a viral vector carrying a functional FMR1 gene that can be used in clinical trials in people with Fragile x syndrome.

Fragile X Research Foundation of Canada partners with FRAXA

We are very pleased to announce that a special funding partnership has been arranged between the Fragile X Research Foundation of Canada and our U.S. counterpart, the FRAXA Research Foundation. Since both foundations are dedicated to funding biomedical research for improved treatment, and ultimately a cure for Fragile X, this funding partnership will allow your donations to go twice as far.

The first grant awarded under this arrangement will go to the laboratory of Dr. Nahum Sonenberg, McGill University, Department of Biochemistry, in support of the project entitled "Aberrant insulin signaling in a mouse model of Fragile X syndrome"

Insulin signaling is known to be dysregulated in diabetes and cancer, and has lately been described to be implicated in cognitive impairments in neurodegenerative disorders such as Alzheimer's disease. Also, dysregulation of insulin signaling might be associated with autism. This team will systematically investigate the impact of insulin signaling on autistic-like behaviors, synaptic plasticity, spine morphology and mRNA translation (protein production) in the mouse model of Fragile X syndrome. Targeting insulin signaling might uncover novel drug treatment options for Fragile X syndrome.



Dr. Nahum Sonenberg (Left) and Dr. Ilse Gantois (Right)

Did you know....

Fragile X encompasses a group of conditions that can affect family members in different ways: Fragile X syndrome (FXS), fragile X-associated tremor/ataxia syndrome (FXTAS), fragile X-associated primary ovarian insufficiency (FXPOI), and other premutation carrier issues.

Project i-Help Internet based help for families of young boys with FXS

Researchers in the U.S. are recruiting families of boys with FXS, ages 2 - 5 years of age, who engage in challenging behavior such as aggression, self-injury or other disruptive behavior to participate in a study evaluating behavioral interventions to decrease child challenging behavior and improve family quality of life.

The study is 18 months long and involves assessment to determine the reason for your child's behavior, parent training and coaching in behavioral intervention sessions during family routines and long term monthly follow up to determine if positive outcomes have maintained.

All sessions take place in the family's home via telehealth technology.

Parents are loaned a laptop, web camera, and bluetooth headset for use during the study.

Participating parents will receive a \$100 gift certificate in appreciation for their participation.

This study has been approved by the University of Oregon Institutional review board (approval #06302015.046) and is funded through the Jerome LeJeune Foundation.

Families can learn more about the study including inclusion criteria by visiting our webpage <https://blogs.uoregon.edu/ihelpinfo/about-2/>

For more information about the study, families or professionals can contact Dr. Machalicek at wmachali@uoregon.edu or 541-346-4404.

Wendy Machalicek, Ph.D., BCBA-D

In Memory

Earlier this year, we received a note from Marge Borne, sharing the sad news that her son, Regg Borne, had passed away.

"It was 1977 when we brought both boys to Vancouver for testing that a doctor who had done her thesis on Fragile X told us that our sons fit into this category. They were then the 2 youngest boys in B.C, and were followed by medical staff and a genetic counselor. We were also part of the FRAXA conference in New Mexico in the 90's.

Being in B.C. we were not always able to take part in some of the research. We were very involved in getting the cluster model going here in Kelowna. 4 men each had their own condo and some form of work to go to during the daytime.

For supper, they would come together in the kitchen apt. where staff had prepared dinner for all of them."

With a mixture of sadness, immense joy and gratitude we said farewell to our beloved son and brother, Regg. He loved to ride around Kelowna using the bus system. During the last several months of his illness a special friend arranged for rides on a double decker bus. How fitting then, that on Monday, April 3, he boarded the 9:20 am bus for heaven.



Regg Borne, October 10, 1972 - April 3, 2017

The gene responsible for Fragile X is called FMR1 and is found on the X chromosome. Everyone has the FMR1 gene on their X chromosome.

Grass Roots Advocacy for Students with Fragile X syndrome

By Barbara Byers:

As parents we all deal with schooling issues for our children. It may be educating a teacher who knows little about FSX, or changing the mind of a principal that says there are no funds to hire the Teaching Assistant that your child needs in order to learn. There are fundamentals that can be attributed to FX kids, such as learning from the whole and not the pieces, learning through imitation, learning in environments that don't offer too many stimulants. Every day you try to get those messages across, knowing in your heart that there would be no "behaviour issues" if only a few accommodations were made for your child. Managing the outside world that interfaces with our kids can be a full time job! While throwing your hands up and yelling might be a quick and easy way to dispel the stress, I encourage you to look into a Committee that every school board in Ontario must operate. That is the "Special Education Advisory Committee".

Each SEAC has 12 seats available for representatives of various Special Needs communities. I have been a member of the Peel District School Board SEAC for 15 years, 14 of those as the Chair. We have representatives for the Blind and Visually Impaired, Deaf and Hard of Hearing, Community Living, Easter Seals, Mental Health, Autism, Learning Disabilities, Gifted, Fetal Alcohol Syndrome, Down Syndrome, Tourette Syndrome, and Fragile X Syndrome. The SEAC is

able to have two community members appointed as well. The school board is required to consult with its SEAC on the annual Special Education Plan and the Spec Ed Budget. Other issues also arise during the 10 month period it operates each year and many boards have parent education events where SEAC members have tables and distribute information. SEAC members can also advise parents on IPRCs and IEPs and can attend with the parents when those meetings are held. SEAC meetings are open to the public and you may wish to attend one to see how they operate. Information on dates and times should be posted on your school board's website.

While large school boards, such as Toronto, Peel, and Halton generally have "full" SEACs, I have found by attending provincial conferences that many places have low participation on their special education committees and are always looking for more members to join. A prospective member has to reside in the community represented by the school board and must be nominated by the group they represent. The FXRFC will gladly nominate you if you wish to participate in a SEAC; contact us with your name, the school board name and address, and the name of the Chair of the Trustees. The insight you will gain, the understanding of the bureaucracy, and the networking with school board personal is invaluable! Good luck and get out there for your children!

Giving Tuesday 2018 is almost here!

Most people know about Black Friday and Cyber Monday ... **but have you heard of Giving Tuesday?**

It's now in Canada and it happens on November 28, 2017.

It is a fairly new Canadian movement for giving and volunteering, taking place each year after Cyber Monday. The "Opening day of the giving season," it is a day where charities, companies and individuals join together to share commitments, rally for a favourite cause and think about others. Giving is a lifelong commitment, and a giving day is a special celebration, a reminder to all of us to think of others and our communities as well as ourselves. Giving Tuesday is all about charity! It's about creating a national movement for giving at the start of the annual holiday season. It celebrates and encourages donations that support charities and non-profit organizations like ours! Of course, we also encourage giving back throughout the year, whether through a monthly donation, or in whatever one time annual donation you can give.

Giving Tuesday is more than a moment, it's a movement.

We ask that you, the families that the Fragile X Research Foundation of Canada was created for, think of the FXRFC on Giving Tuesday and then help spread the word to your networks. Simply go to our main website and click on the Donate Now button. Thank you and we wish everyone a Happy Holiday Season!



The Annual FX Foundation Family Fun Day was a Hit!!

By Jen Williams and Lori Beesley

On June 11th of this year, we held our 14th annual FXRFC Family Fun Day at Variety Village in Scarborough, Ontario. This year's picnic was very successful, with many new families and some of the researchers we fund all in attendance. The weather cooperated and everyone had a good time! As the families mingled and got to know one another, the BBQ was busy cooking up burgers and hot dogs to feed the crowd. The kids blew bubbles, kicked balls, played on the wonderful equipment and special swing, and everyone had fun with the giant parachute - always a crowd favourite!

This year we had a wonderful new donation from COBS Bread of Upper Canada Mall. A very special THANK YOU to Jocelyn Roche, Bakery Manager, for all the buns and some yummy scones! We would also like to give a very special SHOUT OUT to Bill Goodwin of Party City. They have been generously supporting the picnic with an amazing gift card for many years. We use every penny to help to make the picnic not only a success but fun for the families that attend. One of our FX dad's, Ryan Farquhar, always comes through with securing this donation so we'd like to thank him and send along our appreciation.

Of course we have to thank our small but mighty team of volunteers who come out year after year to help. They donate their time, working behind the scenes leading up to the day, arrive early to set up and work tirelessly throughout the event. We could not do this without them and we thank everyone of them most sincerely.

Plans for the 2018 picnic are already in the works. If anyone would like to help, we are always looking for volunteers, especially teenagers who are looking for high school volunteer hours. We always need them the day of! The cost of the picnic is held to a minimum every year due to great donations like to ones mentioned above, but we are always looking for more! We have been told we are losing one of our regular food donations next year due to a change in corporate policy. If you or someone you know works for a company that would like to make a food or goods donation, or perhaps make a corporate "sponsorship" donation, please contact us through the FXRFC head office at 905-453-9366.







Important Information

We are looking for letters, stories, or articles about an Xxx-tra special person!

The FXRFC newsletter often publishes stories about the people that sparked the formation of the Canadian Fragile X Research Foundation and inspires the ongoing growth and commitment of the FXRFC. If you have a story about a child or adult with Fragile X syndrome, please send it to us with a picture of the X-tra Special Person to share with our readers. Tell us a funny or heart-warming story, or share a success and allow it to inspire others. We love to hear what is happening with our Fragile X families across Canada, so please consider sharing your story and letting others share in a success!

Just e-mail us at newsletter@fragilexcanada.ca

Fundraising for the FXRFC

The Fragile X Research Foundation of Canada is a 100% volunteer organization and we are always in need of fundraising dollars! How much you contribute and how much time you contribute to a fundraiser differs from person to person. We value all of our contributors, however they choose to raise funds and awareness. Whether you're a family member of someone with Fragile X syndrome, a friend, or just a community-

mindful individual, we welcome your contribution of time, energy and dollars. If you are interested in fundraising with the Foundation please contact us or go to the Volunteering page on the How to Help section of the FXRFC website. You can also contact our National Fundraising Coordinators, Ian Shearer and Jennifer Williams, by sending an email to fundraising@fragilexcanada.ca or volunteer@fragilexcanada.ca

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New PDF ONLY version of the newsletter!

This newsletter is produced 3 times a year by the Fragile X Research Foundation of Canada and emailed to our supporters. You can also view/download a PDF of the newsletter on our website at www.fragilexcanada.ca by clicking on Resources. If you do not receive our newsletter regularly or would like us to email a copy to someone you know, send an email with a complete email address to newsletter@fragilexcanada.ca

The Fragile X Research Foundation of Canada (FXRFC) is a national, nonprofit,

tax-exempt charitable organization run by parents of children with Fragile X syndrome and volunteer professionals. Permission is granted to reproduce or distribute this newsletter for non-commercial purposes. We accept donations on-line through our website at www.fragilexcanada.ca or by mail or phone at our National Headquarters in Brampton, Ontario.

Charitable Registration Number:
88643 3762 RR0001.

"Like" Us on Facebook!

The Canadian Fragile X Research Foundation is on **Facebook!** Please go to our page and "Like Us"! In the months to come, we are hoping our page will become a great forum for our Fragile X families. What a great way to connect with each other, share ideas and info. and support one another. Feeling isolated? Maybe there is another Fragile X family near by... Check out the page and stay in touch!

www.fragilexcanada.ca

Please keep checking the new FXRFC website for upcoming events. If you have an event planned in your area, let us know and we'll include it on the site and in our next expressions newsletter issue.

We frequently update the Fragile X Research Foundation of Canada website with new information. Check out our What's new or How close are we to a treatment sections regularly to get updates on the FXRFC.

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