



# FXRFC Newsletter

News for Friends of the Fragile X Research Foundation of Canada

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## FXRFC Supports Orphan Drug Policy

The Fragile X Research Foundation of Canada recently conducted a mailing campaign to support The Canadian Organization for Rare Disorders (CORD) in advocating for the creation of a Canadian Orphan Drug Policy (ODP). An orphan disease is one that affects people at a frequency of one in two thousand (1/2000). The designation of an “orphan disease” varies depending on the respective country, ranging from 1/9,090 in Australia to 1/1,333 in the United States. In Japan and Europe the figure is set at 1/2,000. Canada is the only developed country in the world without an ODP.

Today there are drugs and treatments available for some of the hundreds of orphan diseases that have been described, however Canadian patients cannot access these drugs because individuals afflicted with these rare diseases are not covered under the Canada Health Act. Canada requires a national Orphan Drug Policy to ensure that patients with orphan diseases are treated equally to those with more common illnesses.

The significance of this Canadian shortcoming is highlighted by the fact that there are drugs available in the U.S. that are being used to treat the symptoms of Fragile X, but are not available in Canada. One example is a drug called Abilify (Aripiprazole), which has been approved for treatment of schizophrenia for several years in the U.S. It has recently been used very effectively to treat patients with Fragile X. The drug can decrease anxiety, improve concentration and control aggressive outbursts in Fragile X patients. When we contacted the Canadian branch of the pharmaceutical company that manufactures Abilify, we were informed that there are no plans to make the drug available in Canada, simply because it is not part of their marketing plan and there is no economic motivation to seek market approval in Canada. If a drug that has already been used safely in other countries cannot be obtained in Canada, how much more difficult will it be to bring newly developed treatments for Fragile X to Canada?

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FXRFC is a nonprofit, tax-exempt charity run by parents and volunteer professionals. Fragile X syndrome is the most common form of inherited mental impairment and developmental disabilities, affecting approximately 1 in 2000 males and 1 in 4000 females. FXRFC's goal is to accelerate research aimed at the treatment of Fragile X, by direct funding of promising research projects and by raising awareness of this disorder.

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The answer to this problem is the creation of a national Orphan Drug Policy. The Fragile X Research Foundation of Canada supports CORD's efforts to help create a Canadian Orphan Drug Policy (ODP) that has guidelines and opportunities to provide/develop/offer treatments for rare disorders in Canada. The Orphan Drug Policy (ODP) would develop a social program that will provide funding to enable individuals affected by rare disorders to obtain access to those treatments.

Health Canada is currently working on draft legislation for a Canadian Orphan Drug Policy. The Federal, Provincial and Territorial Health Minister(s), along with the deputy ministers met in October, 2005 and a Canadian Orphan Drug Policy was on the agenda. The FXRFC took this opportunity to mount a letter writing campaign to all Federal, Provincial and Territorial ministers to support CORD's efforts to influence the development of a Canadian Orphan Drug Policy. For more information about the ODP you can go to CORD's website at [www.cord.ca](http://www.cord.ca).

How can you help? Write to your local MP, and Provincial and Federal Health Ministers. The names and addresses of the government ministers, along with a suggested letter will be posted on the FXRFC website at [www.fragile-x.ca](http://www.fragile-x.ca).

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## Research Update

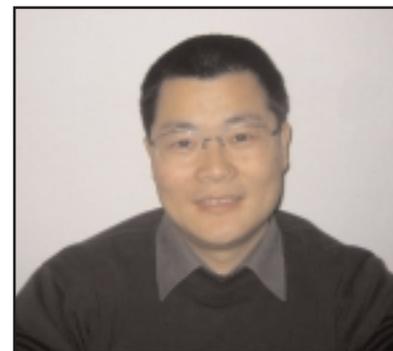
*The following article is an update on a research project funded by the FXRFC. The postdoctoral fellowship, worth \$40,000.00, was awarded to Dr. Min-gao Zhao in Dr. Zhuo's lab at the University of Toronto. His findings were recently published in The Journal of Neuroscience, August 10, 2005.*

### **Deficits in Trace Fear Memory and Long-Term Potentiation in a Mouse Model for Fragile X Syndrome**



*Min Zhuo Ph.D. (Principal Investigator)  
Department of Physiology, Faculty of  
Medicine, University of Toronto*

The fragile X syndrome is a common genetically inherited form of mental retardation, causing cognitive impairment, macroorchidism and behaviour abnormalities. Recent genetic studies suggest that it is caused by the loss of an important protein called the fragile X mental retardation protein (FMRP). FMRP can regulate new protein synthesis within synapses, the key neuronal structures for brain functions such as learning and memory. It is generally believed that the brain needs to synthesize



*Min-gao Zhao Ph.D.  
(Postdoctoral fellow)*

new proteins used for synaptic functions and memory storage. The loss of FMRP will result in the misregulation of local protein synthesis, and thus impairment of brain functions.

This proposal is the first study to systematically map synaptic plasticity in three main memory-related regions of the brain: the hippocampus, amygdala, and anterior cingulate cortex (ACC).



Synaptic plasticity refers to the ability of neurons in the brain to continuously change their synaptic connections with each other in the process of learning. The two main types of long-lasting synaptic plasticity, known as Long-term Potentiation (LTP) and Long-term Depression (LTD), are believed to be involved in learning and memory.

In previous studies, we demonstrated that FMR1 knockout (KO) mice show a defect in trace fear memory, but did not differ from normal wild type mice in terms of activity level, anxiety, or pain sensitivity. Trace fear memory requires the activity of the anterior cingulate cortex (ACC) and is sensitive to attention-distracting stimuli. Because many children with Fragile X are diagnosed with attention deficit/hyperactivity disorder (ADHD) (Hagerman et al., 1996; Cornish et al., 2004), we hypothesized that this form of associative learning may be disrupted in FMR1 KO mice and alterations in synaptic plasticity within the ACC may underlie any observed deficits.

A technique known as “Whole-cell path clamp recordings” in the ACC was used to measure electrical activity in the neurons and showed that long-term potentiation (LTP) was completely abolished in this part of the brain in the knockout mice. A similar decrease in LTP was found in the lateral amygdala, another brain structure implicated in fear memory. No significant changes were found in short-term synaptic plasticity, or regular, ongoing basal synaptic transmission. This suggests that synaptic plasticity in the ACC and amygdala of *FMR1* KO mice play an important role in causing the behavioural changes similar to the symptoms of Fragile X syndrome.

In a proposed study, we will do further detailed mapping of the synaptic responses in KO mice. Second, we will examine the effects of potential drugs on synaptic plasticity to see if drug treatments rescue changes in KO mice. Finally, behavioral memory experiments will be performed to determine if drug treatments rescue the behavioral defects in memory. Our proposed studies will help us understand the mechanism of cognitive impairment, thereby providing possible new molecular targets to treat patients.

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### *Think Fragile X when doing your Holiday shopping...*

*With December right around the corner, we know that some busy families are already turning their thoughts to holiday shopping. At the FXRFC, we would like to suggest you consider making a donation in someone special's name as an alternative to buying a gift.*

*Some of our families do this on an annual basis, with both the sender and the recipient getting a letter from the Foundation, making them aware that the donation has been received. If you would like to include a small gift along with the donation, may we suggest a FXRFC cook book that you could inscribe with a personal message. All the compiled recipes were sent in by friends and families touched by Fragile X. Cookbooks are available for \$10 by contacting the FXRFC headquarters at (905) 453-9366 or by e-mail at: [fxrfc@attglobal.net](mailto:fxrfc@attglobal.net).*

*Happy Holidays!*

## Wine Festival a Tremendous Success

The end of September 2005 was a very busy time for outreach and fundraising for the FXRFC, thanks to **Garth and Lesley Buchko** of Winnipeg, Manitoba. Dr. Carlo Paribello and his wife Barbara Byers, flew out to Winnipeg on September 22 and were greeted by Garth and Lesley, the parents of 15 year old Matthew, who has Fragile X Syndrome. That evening Garth, who is the General Manager of **CJOB Radio** in Winnipeg, had arranged for Dr. Paribello to be interviewed on **CanWest Global** television by broadcaster **Charles Adler**. The interview was shown that evening and helped bring awareness of Fragile X Syndrome to the thousands of TV viewers who tune into Mr. Adler's thought provoking interviews nightly.

The next morning Mr. Buchko, thanks to the help of **Dr. Chudley** in Winnipeg, brought together over 30 families from all over Manitoba who have loved ones that have Fragile X Syndrome. Many had been unaware of the Foundation and they were delighted to find a group that understands their struggles and is funding groundbreaking research for a treatment and cure. As part of the seminar given by Dr. Paribello, the group also heard a presentation by **Dr. Tammy Ivanco**, an FXRFC funded researcher with the University of Manitoba. Dr. Ivanco brought along a microscope through which participants could view an actual FX dendrite from a mouse nerve cell. Later on in the day, Dr. Paribello was taken on an in depth tour of Dr. Ivanco's laboratory seeing first hand the exciting work that the FXRFC is funding.

Friday afternoon, Dr. Paribello was on air at CJOB, again being interviewed by Charles Adler, for a national radio presentation. The program is already generating contacts and interest from across Canada and we are tremendously grateful to Garth Buchko for affording the Foundation this unique outreach opportunity.

The main event was a spectacular evening gala arranged by Lesley and Garth Buchko at the St. Charles Country Club in Winnipeg. Titled "Somewhere Over the Rainbow Dreams Do Come True!" the event featured wine tasting courtesy of *Mark Anthony Brands* and *Kenaston Wine Market*, beer tasting courtesy of *Wett Sates*, and martini tasting courtesy of *Maxxium Canada*. A host of generous donators created a dazzling Silent Auction table that



*Dr. Paribello & Garth Buchko*



*Mathew Buchko, his Aunt Susan (centre) and mother Lesley Buchko (right)*

featured Winnipeg Symphony tickets, fine art, trips to Montreal and Toronto, sports tickets, and golf memorabilia.

The grand prize was a trip for two to Vienna on Austrian Airlines. The night ended all too soon, but with an amazing total of **\$30,000.00** raised for Fragile X Research!

*The FXRFC sends a warm and sincere thank you to Garth & Lesley!*

## IFDS Golf Tournament

On August 4, 2005, **International Financial Data Services** held its annual Charity Golf Tournament at the magnificent Blue Springs Golf Club in Acton, Ontario and The Fragile X Research Foundation was honoured to be the recipient of the proceeds of this year's event.

International Financial holds an annual client golf tournament in support of charities that are important to its staff, shareholders, business partners, and clients. **Frank Zitella**, an employee of IFDS and father of a boy with Fragile X Syndrome, was thrilled when the organizing committee for this event approached him and offered to support Fragile X research.



*Frank Zitella gets ready to hit the links*

IFDS clients were invited to play and support the charity by betting on their golfing abilities on holes throughout the course and by getting involved in the silent auction. Suppliers and affiliates sponsored and provided prizes and silent auction items for the players. By the end of the day, the tournament had raised **\$51,000.00** for the Fragile X Research Foundation of Canada!

*The FXRFC would like to thank Frank, the IFDS staff, business partners, clients and sponsors for making this event such a tremendous success!*

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## The Fragile X Awareness Wristbands are Here!

*Our bright green silicone wristbands say "Support Fragile X Research" and show the FXRFC logo. It's a great way to raise public awareness about Fragile X and raise money for research at the same time! Order 1-4 wristbands at \$3 each or save, and order 5 or more wristbands at \$2 each. Try selling them at work as a fundraiser, or what a great gift idea for teachers, babysitters, friends and family.*

*Just call the FXRFC headquarters at (905) 453-9366 and order yours today!*

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## FXRFC Bulletin Board

- Need a Musical Act for your Fragile X Fundraising event? John Henderson of Grand Stand Entertainment has volunteered to host benefit concerts and provide musical entertainment for Fragile X Fundraising events held in the "Golden Horseshoe" area of Ontario. If you would like to plan such an event, you can contact him at: office: 905-468-0601 cell: 905-730-9911.
- Fragile X Benefit Concert. The "High Energy Rock'n Roll Band", Hammersmith Gritt, will perform a Benefit Concert in support of research to find a cure for Fragile X Syndrome  
Sat. Nov 5th @ The Irish Centre - 20 Strathearne Ave., Brampton, Ontario  
We hope to see you out for this worthy cause. All ticket proceeds will go to Fragile X research. Hammersmith Gritt is honoured to be part of this event. Tickets \$10. Call 416-760-6542 for info.

## City cop tests his endurance

By Terri Saunders

Reprinted from the Friday, Sept. 9th, 2005 edition of the Cornwall Standard-Freeholder

### It's all for a good cause, raising money for Fragile X Foundation



**BEHIND THE WHEEL** Tony Pettinella, back left, and Denis Lafave, back right, check out the interior of a stripped city police cruiser with Const. Dave MacLean at the wheel. MacLean will be hitting the track at the Cornwall Motor Speedway Sunday afternoon to raise money for charity.

Const. Dave MacLean doesn't get a lot of respect at the Cornwall Motor Speedway - at least not right away. "When they announce me, sure there are cheers, but there are a lot of boos at the same time," said the Cornwall Community Police Service officer on Thursday. "When they know I'm a police officer, and they see the cruiser, I certainly become a target."

But it's all in good fun, and it's all for a good cause. For the second year, MacLean will be behind the wheel of a stripped-down police cruiser to take part in the Fireball Enduro being held at the track on Sunday at 2 p.m.

Hoping to make it through to the end of the 50-lap competition in one piece, MacLean said his hopes are also high when it comes to the amount of money he'd like to raise for the Fragile X Research Foundation of Canada.

"People can sponsor me in the race, and I was trying to decide which charity to raise money for," said MacLean. "I thought Fragile X was the perfect choice."

Fragile X is a disease which can cause an intellectual impairment, attention deficit disorder, hyperactivity, anxiety and unstable moods, autistic-like behaviour, physical attributes such as a long face, large ears, flat feet and hyperextensible fingers, as well as epileptic seizures. The name comes from the fact the disease is carried on the X chromosome.

One of MacLean's friends, Denis Lafave, knows all too well about the disease. Both his sons, Brandon, 10, and Benjamin, 7, were diagnosed with the disease at young ages, and with his wife Jean, the family was thrilled when they heard MacLean wanted to raise money for a cause so close to their hearts.

"Dave gets work done here," said Lafave, who owns Direct Muffler Brake and Auto Services, on Pitt Street. "We know each other because of the business side of things, but we're also friends, we sponsor Dave's ball team and we talked about it and he said he wanted to raise money for Fragile X."

On Sunday, MacLean will hit the track in the stripped down Caprice, powered by a Corvette LT motor, and hopes he makes it through to the end.

"Last year, I got through 25 laps myself before the race was officially over, so that was great," said MacLean, who added the car will be auctioned off after the race will all proceeds going to the Fragile X Foundation.

"I'm hoping to do even better this time around."

*Const. Dave MacLean did a great job the FXRFC. Thank you for the \$1,400 donation.*

*Good Luck next year!*

## **XXXX-tra Special Person!**

The FXRFC Newsletter often publishes stories about the people that have sparked the formation and inspired the growth of the Fragile X Research Foundation of Canada. If you have a story about a child or adult with Fragile X, please send it to us and we will share it and a picture of the X-tra Special Person with our readers. Tell us a funny or heart-warming story, or share a success.

This newsletter's X-tra Special person is 6-year-old Tyler. The following poem, entitled "My Special Brother" was sent in by Tyler's mother, Elizabeth Burgie. She wrote this from the loving eyes of Tyler's younger brother Andrew and in anticipation of his new baby brother due in November.



*Tyler*

### **MY SPECIAL BROTHER**

*My brother Tyler is special to me,  
Unlike many others he's different you see.*

*He's quiet and gentle and loves to hug,  
He plays with your hair when you need some love.*

*He loves trains, books and movies too,  
Watching trucks drive by and in the pool.*

*I try to play with him but he moves away,  
Sometimes I'm lucky and he will stay.*

*He usually watches me play with our toys,  
And will plug his ears if I make too much noise.*

*He goes to class in a regular school,  
Has a teacher's aid that is really cool.*

*He gets all kinds of therapies and speech too,  
That's hard to say if you're fewer than two.*

*He may not talk but he has lots to say,  
Using signs and pictures he pulls us his way.*

*When he's very happy he squeals and smiles,  
And waves his arms in a fast flapping style.*

*He's beautiful and kind and a wonderful boy,  
Everyone loves him; he brings all such joy.*

*He's never angry or never bad,  
He's always happy and never sad.*

*He has Fragile X; it's from our mother,  
But I love him so because he is my brother.*

## **FXRFC Support Groups - We are Growing!**

We are very interested in starting support groups in the other provinces and territories and will provide assistance to anyone able to help out. If you would like to network with other parents of children with Fragile X, or if you would like to volunteer for fundraising events in your area, please contact any of the people below.

### ***National Headquarters***

167 Queen St. W.  
Brampton, Ontario, Canada  
L6Y 1M5  
Phone: (905) 453-9366  
E-mail: [fxrfc@attglobal.net](mailto:fxrfc@attglobal.net)

### ***Quebec Support Group***

Evelyn Lockett  
32 Lakeview  
Baie d'Urfe, Quebec  
H9X 3B1  
Phone: (514) 457-3284  
E-mail:  
[evelyn\\_lockett@hotmail.com](mailto:evelyn_lockett@hotmail.com)

### ***Ontario***

#### ***Kitchener-Waterloo Fragile X Support Group***

Teresa Burch, Co-ordinator  
62 Corfield Drive, Kitchener,  
Ontario N2A 3W9  
Phone: (519) 894-3689  
Fax: (519) 894-6333

### ***British Columbia Support Group***

Pam Winthrope  
7968-112A Street  
Delta, BC V4C 4Y6  
Phone: (604) 590-6822  
E-mail: [pwinthrope@hotmail.com](mailto:pwinthrope@hotmail.com)  
Web:  
<http://fragilex.bc.tripod.com/fragilex.bc/>



*Please become a Friend of The FXRFC  
in supporting research aimed at treatment  
for Fragile X. Friends of the FXRFC  
receive this quarterly newsletter and are  
welcome to participate as active volunteers.*

## **FXRFC Newsletter**

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The Fragile X Research Foundation of Canada (FXRFC) is a national, nonprofit, tax-exempt organization; charitable # 88643 3762 RR0001. You can become a friend of the FXRFC for a tax-deductible donation of \$35 or more per year. The FXRFC is a 100% volunteer organization run by parents and professionals - which means more of your donation goes directly towards research. We now accept VISA and Mastercard payments; simply phone (905) 453-9366 and pass on the details. You may send your cheque or money order to:

**The Fragile X Research Foundation of Canada**  
167 Queen St. W., Brampton, Ontario, Canada L6Y 1M5  
*Come visit our site on the web at: <http://www.fragile-x.ca>*