



FXRFC Newsletter

News for Friends of the Fragile X Research Foundation of Canada

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Stem Cell Research Generates an Important Discovery for Fragile X Syndrome

Dr. Laurie Doering, Associate Professor, Department of Pathology and Molecular Medicine.



The FMR1 gene in Fragile X leads to inactivation of the gene product, known as the Fragile X Mental Retardation Protein (FMRP). The Fragile X research community continues to provide new insights and knowledge about the function of FMRP. Increasing evidence suggests that FMRP plays a role in “synaptic plasticity” – a process that influences the functional sites (synapses) in the brain. Until now, FMRP has been thought to be confined mainly to neurons in the nervous system. Recent experiments by **Laura Pacey** (Ph.D. student in Dr. Doering’s laboratory) have led to the finding of FMRP in another major class of cells in the brain known as glial cells. Performing experiments with adult brain stem cells, Laura observed FMRP in developing astrocytes (a sub-class of glial cells).

Dr. Laurie Doering

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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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This finding is important because astrocytes play important roles in the development of the nervous system and in the maintenance of normal communication between neurons. Astrocytes are essential for normal activity in the brain.

The absence of FMRP in astrocytes may contribute to the abnormal neuronal structures seen in the brains of Fragile X patients. The results of this research have strong implications and can open new streams of research that center on glial cells as vital players in the pathogenesis of Fragile X.

The findings were published on-line September 06/07 in the prestigious journal “Glia”.

This research is supported by the Fragile X Research Foundation of Canada.

Fragile X Clinic – Providing Assessments to Canadian Residents of all Provinces and Territories

Medical assessments at the Fragile X Clinic at Surrey Place Centre in Toronto are now available to all Canadian residents with a valid health card from their home province. The clinic serves as a centre to assess and treat patients of all ages who have Fragile X syndrome.

It is located in the heart of downtown Toronto, and is the first dedicated Fragile X Clinic in Canada. It provides affected patients and their families with access to physicians with the expertise and knowledge to treat this neuropsychiatric disorder.

The Fragile X Clinic is also in the process of preparing for clinical trials with new drugs that enhance learning, memory, and cognitive functioning and will therefore put Surrey Place Centre in a position to make a global impact in the field of neuroscience and autism.

These services are offered free of charge to their clients because Surrey Place Centre receives core funding through the Ministry of Community, Family and Children’s Services. The Surrey Place Centre Foundation also raises funds from individuals, corporations, and foundations for additional support. It is affiliated with the University of Toronto, York University, the University of Guelph and many other teaching institutions. It is accredited with the Canadian Council on Health Services Accreditation.

All patients will be assessed **Dr. Carlo Paribello** M.S.M, M.D., Family Physician, President, Medical Director, Fragile X Research Foundation of Canada and **Leeping Tao**, RN, MN-ACNP, Nurse Practitioner, Surrey Place Centre. If required, patients can be referred to allied health professionals at Surrey Place Centre and Toronto General Hospital.

The recommendations that arise from these assessments will then be given to the patient’s family members, caregivers or workers for implementation by their local healthcare providers.

For further information about the clinic and for patient booking, please contact the office of Dr. Carlo Paribello at (905) 453-9366 or e-mail your request to fxrfc@on.aibn.com.



Letter from the Millar's - Brady's Golf Tournament



“We are writing today to tell you of our successful golf tournament to raise funds for your charity.

Brady's grandfather, **Tony Mercier**, started organizing the event in August for a September 17th tournament date. He got tremendous responses from people wanting to play, come for the dinner and donate prizes and money. Within 3 weeks we had 52 golfers and 66 people attending the dinner. It took place at The Oaks of Cobden in the Ottawa Valley area. It was a beautiful, sunny day for golf and we had a BBQ steak dinner afterwards.

Everybody went home with at least one prize. We are hoping to continue this tradition for many years to come and help raise awareness of Fragile X in the area. Two small local newspapers are writing stories of our event as well. The final total raised was \$6,460.00! We know our 3½ year old son Brady will benefit greatly from your research.

Thank you for being here for us and so many other families as a light of hope.

Sincerely,

Carla and Andrew Millar”

The Dave Robinson Memorial Charity Golf Tournament to Benefit The Fragile X Research Foundation Of Canada

By Kim Fitzgerald, Peterborough, Ontario



Brian McGregor - putting contest

On Saturday September 15, 2007 a golf tournament was held in memory of my brother **David Robinson**. My brother passed away on November 22, 2006 and it has been a very difficult time for our family and friends. This day in his memory was exactly what we needed, time to share our memories of him.

We ended the day with some drinks, a great dinner and prizes. We were able to raise approximately \$2,900 for the Fragile X Research Foundation. Thank you to Dr. Paribello for joining us at dinner and sharing his knowledge of the

research that is taking place as well as his own personal stories about raising his two sons. My son Brian, who has Fragile X, had a great time hanging out with his Uncle's buddies. Kayla, my daughter, was able to share stories and laughter with everyone that attended.

My brother is missed by all who knew him.



Dr. Paribello, Kim Fitzgerald (centre) and Brian McGregor and Family



Help Us Build a Resource List

Dr. Myra Sourkes, M.D.

I am a Fragile X parent and member of the board of the FXRFC. I am in the process of compiling an updated resource list of organizations and programs that provide leisure and therapeutic activities for children with special needs, including Fragile X Syndrome. The current list is based on my own experiences and those of other Board Members and friends; it is neither official nor exhaustive. Many of the programs of which I am aware are situated in the Toronto area, where I live, but I am sure that there are other excellent programs across the country.

I am asking you, the readers of the newsletter, to let me know about programs that have helped you and your family, wherever you live. Please send your tips to the FXRFC at 167 Queen St. W., Brampton ON L6Y 1M5 or fxrfc@on.aibn.com.

The list will appear in a subsequent newsletter and eventually will be posted on our website.

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 cookbook 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. Happy Holidays!



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 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.

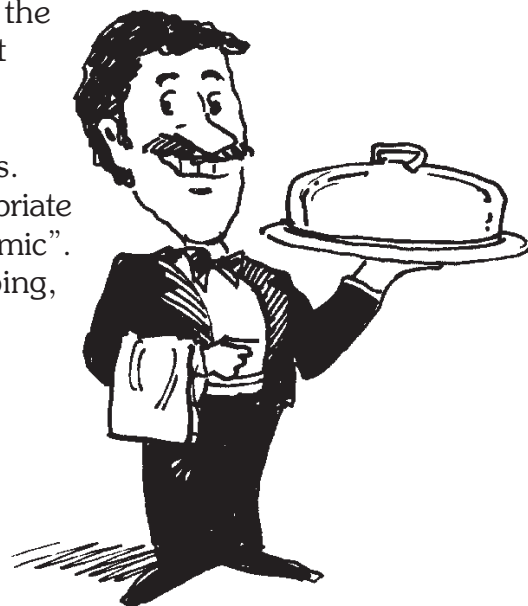
Fine Dining with the Mob

*By Barbara Byers,
Legal counsel, FXRFC Board Member*

There was a time when I wouldn't have dreamed of taking my kids out to a restaurant. Well maybe I would take them to McDonald's, if pressed. There was a huge play area there where kids could dive into the plastic balls, scramble up the staggered staircase, crawl through the mile high tubes, and slide down the spiral slide, all the while yelping and hooting with the best of them. Some how the cacophony of McDonald's Playland wasn't the same type of noise encountered in the No Frills grocery store. That kind of noise was like an electric jolt to my son Ben. It seemed low grade to me, but I knew that he heard the fluorescent lights buzzing, the continuous beep of the cashiers' scanners, the din of the Muzak, punctuated by the occasional piercing screech of a toddler denied a chocolate bar. Week after shopping week, I expected that the conditioning would acclimatized him to the racket; not so. One day, when he was around 5 or 6 years old, after consistently asking me when we were going home and getting more and more anxious, he stopped dead in his tracks. He bent over at the waist, rapidly stomping on the spot, fists clenched by his ears, squinty eyed and screaming. Before I could give him a centering shoulder squeeze, he dropped spread eagle on the floor and stared blankly at the ceiling. I got down on my knees and gently whispered in his ear that we were going home. I left the cart, full of groceries, in the middle of the aisle and walked with him out to the car.

So how, you ask, did I think I could ever get him to sit in a noisy restaurant? The answer is the local Chinese buffet. It is a perfect place to take the sensory challenged to eat! We go to a wonderful restaurant made up of cavernous dining rooms that feature fish tanks and caged birds to entertain you while you eat. But the part that "works" for Ben, is the ability to keep getting up and down to select from the buffet. There is no commitment to staying at the table or having to wait until everyone is served before starting to eat. The heady independence of choosing what to eat and how much of it, plus being able to change your mind about your fondness for calamari rings after you've been told about the dessert options, makes for a very successful outing. It is just a whole lot of fun and he doesn't become burdened by the onslaught of noise that surrounds him. The only FX issue is that I have to work at stifling his gift for imitation when the waiter asks for our order with his accent intact. Ben will light up and start laughing, suddenly "playing Chinese waiter" to his brothers. We have to repress our own laughter and tell him it's not appropriate to imitate the man. "Yes", he'll respond, rolling his eyes, "don't mimic". Then he merrily trots off to get more egg rolls amidst the bumping, calling, and clanging of other patrons.

We aren't contemplating Susur or Jump yet, but I feel that Ben's comfort level with how busy restaurants operate means we can start venturing to Montana's and Swiss Chalet. We have come a long way from the Kid's Happy Meal.



FXRFC Support Groups

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.

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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, ON, Canada L6Y 1M5
Come visit our site on the web at: <http://www.fragile-x.ca>