



The National **Fragile X** Foundation

quarterly

A Journal for Families and Professionals

Inside:

■ **COLUMNS**

President's Notes	1
<i>A Time of Transition</i>	
From the Executive Director	3
<i>Volunteers Make Our World Go 'Round</i>	
Public Policy Update	5
<i>Riding Out the D.C. Political Roller Coaster</i>	

■ **FRAGILE X SYNDROME (FXS)**

Braden on Behavior	10
<i>Get Out While the Getting Is Good</i>	
Brain Imaging Studies Open New Frontier in Fragile X Research	14
Special Topics Pamphlet	18
<i>Fragile X Testing: Facts for Patients and Health Care Providers</i>	
Therapy In Action	20
<i>A Primer on Sensory Integration</i>	

■ **FRAGILE X-ASSOCIATED TREMOR/ATAXIA SYNDROME (FXTAS)**

From Uganda, With Support	8
The Five Most Common Questions About FXTAS	9

■ **FRAGILE X-ASSOCIATED PRIMARY OVARIAN INSUFFICIENCY (FXPOI)**

From POF to POI: Evolution of a Term	22
---	----

■ **REPORTS AND ANNOUNCEMENTS**

Parent's Forum	7
<i>A Deborah Kwan Appreciation</i>	
When Two Became One	12
<i>Cover Story</i>	
What You Can Find at FragileX.org	15
<i>Conferences</i>	
NFXF-Sponsored Research Report	16
NFXF-Sponsored Student Research Report	17
Focus on Fundraising	26
<i>A Season of Overflowing Generosity</i>	
Focus on Volunteers	28
<i>Looking Back on a Volunteering Year</i>	

July 23-27, 2008	
Conference Information	23
Getting to St. Louis	24
Call for Abstracts	25



FRAGILE X CONFERENCE

July 23-27, 2008 © The National Fragile X Foundation

Merger in the FX Family

Landmark Accord Joins NFXF and Conquer Fragile X Foundation

Page 12



Issue 29 ■ December 2007

What Is Fragile X?

The term “Fragile X” represents a group of genetic conditions that have a variety of impacts on affected individuals and their families. The conditions include:

- Fragile X syndrome (FXS)—Caused by the *full mutation* of the Fragile X gene, FXS is the most common cause of inherited mental impairment. Its effects range from learning disabilities to severe mental retardation and autism. Symptoms often include unique physical characteristics, behavioral disorders, and delays in speech and language development.

- Fragile X-associated tremor/ataxia syndrome (FXTAS)—A condition affecting some male (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)—A condition affecting some female carriers of the *premutation*.

Fragile X can be passed on in a family by individuals with no apparent sign of the condition. In some families, multiple generations are affected, while in others, it may cause problems in only one person.

Since 1984, The National Fragile X Foundation (NFXF) has been helping individuals with Fragile X, their families, and the professionals who work with them. As research into Fragile X continues, our understanding of who it affects and how it affects them will grow. The NFXF is committed to: 1) supporting and funding all efforts that will increase awareness, 2) improving education, 3) advancing research toward improved treatments and an ultimate cure, and 4) keeping the Fragile X community always well-informed about the progress of these efforts.

The *Foundation Quarterly* is published four times annually by The National Fragile X Foundation. The journal is distributed to members and others by request. Please contact the foundation regarding content or distribution.

Editors: Robert Miller Jeffrey Cohen
Andrew Hidas

Contributing Editor: Linda Sorensen

Design: Karen Ashton

The National **Fragile X** Foundation

PO Box 37 ■ Walnut Creek, California 94597

Phone: (925) 938-9300

Email: NATLFX@FragileX.org

Toll free: (800) 688-8765

Web: www.FragileX.org

Fax: (925) 938-9315

The NFXF Staff:

Robert Miller
Executive Director

Sam Jacob
Assistant Shipping Clerk

Liane Abrams, MS, CGC
Genetics Specialist

Teddy Palmer
Database Specialist

Phil Campbell
Administrative & Program Assistant

Linda Sorensen, MS
Development & Membership Coordinator

Dolores Finn
Bookkeeper

David Salomon
Office Tech

Services offered:

Telephone consultation—*free*

Basic informational packet—*free*

Educational resources (books, videotapes, CD)—*fee*

Local, national, and international conference sponsorship

Referral to medical, genetic and support services—*free*

Educational advocacy

Legislative advocacy

Research grants

Membership with quarterly journal—*fee*

Services offered to:

Individuals, families, professionals, institutions, and students involved with or impacted by Fragile X

Service Referral: Self or professional

Eligibility: All

Service Area: National and international

Dues-Paying Members: Over 1,100

Annual Contacts: 20,000

Website Visits: Over 300,000 annually

Founded: 1984 as a public non-profit 501(c)(3) charitable organization

Federal Tax ID Number: 84-0960471

Funding: Individual contributions

Mission Statement

The National Fragile X Foundation unites the Fragile X community to:

- Enrich lives through educational and emotional support
- Promote public and professional awareness
- Advance research toward improved treatments and a cure for Fragile X.

Cover photo: Steve McKee, outgoing president of the NFXF Board of Directors, with son Nick (top), Harris Hollin, chair of the Conquer Fragile X Foundation, with grandson Matthew. See story on page 12.

PRESIDENT'S NOTES

Steve McKee



A Time of Transition

This is my last column as your president. Effective January 1, 2008, I will step down from this role after two years. It has truly been a pleasure to serve the foundation and the Fragile X community.

I am also delighted to introduce your president-elect, Dr. Don Bailey. Of course, Don really does not need an introduction. He and his wife Pam have been long-term supporters of the foundation. Most recently they co-chaired a fundraising event in their home state of North Carolina, and Don just completed serving as co-chair of our Scientific and Clinical Advisory Committee. Don brings a unique perspective as both a parent and researcher, along with a well-deserved reputation as a true leader in the Fragile X community. I could not think of a better person to lead the foundation at this time, and I look forward to supporting Don in his new role.

I am very proud of what our foundation has been able to accomplish in the last two years. We have continued to serve the Fragile X community in the same ways we traditionally have—providing support, funding research, advocating for increased federal funding, and producing the world's leading international Fragile X conference. During this time, your foundation has also successfully launched a number of major new initiatives, including:

- **Clinics Consortium**—We are actively bringing together the country's Fragile X clinics to support research trials, collaboration, best practices and expansion of services to families.
- **Adults Living with Intellectual Disabilities**—We have initiated a national dialogue on how to impact legislation that will provide a continuum of services to adults.
- **A Broader Mission**—The foundation has broadened the community we serve, from those impacted by fragile X syndrome to the entire family of fragile X-related

disorders, including fragile X-associated tremor/ataxia syndrome (FXTAS) and fragile X-associated primary ovarian insufficiency (FXPOI).

- **Resource Groups Extension**—The foundation has continued to support the increased reach of our national network of resource groups thorough collaborative efforts such as our annual leadership forum.

As notable as these accomplishments have been, perhaps our most far-reaching success over the last two years is the increased impact of our board.


I am especially pleased to announce that effective November 1 of this year, The National Fragile X Foundation merged with Conquer Fragile X Foundation (CFXF), a leading international nonprofit that provides research grants to scientists studying Fragile X (see story on page 12). This merger has been several months in the making. While many have played a role in making it happen, I want to especially acknowledge Harris Hollin, the

founder of CFXF. Harris has demonstrated a unique vision in creating CFXF, making it a major force in Fragile X research around the globe, and now in helping to orchestrate the merging of our organizations for the greater good of the Fragile X community. Boardmember Jeffrey Cohen and Executive Director Robert Miller also spent countless hours working out the details of this arrangement. We are truly excited about the opportunities it presents.

As notable as these accomplishments have been, perhaps our most far-reaching success over the last two years (though not always visible) is the increased impact of our board.

continued on following page →

Our newest boardmembers have made an immediate, and, I believe, lasting impact on the foundation, and have challenged the rest of us to reexamine what it means to be a leader and how to serve the Fragile X community. At a time when it would be easy for our board to be satisfied with the terrific accomplishments of the past, we are clearly focused on the challenges and opportunities of the future.

I thank all of you for your support over the years, and I look forward to working with you in creating the future we want for our families and others affected by Fragile X. 

Steve McKee is the outgoing president of The National Fragile X Foundation Board of Directors.

Email: s.mckee@FragileX.org

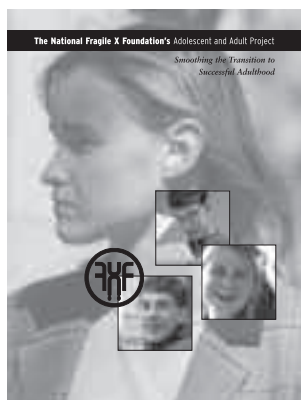
Welcome NFXF President-Elect Don Bailey!

I attended my first International Fragile X Conference in 1994. It was in Albuquerque, New Mexico. I remember being overwhelmed by the science and amazed at the fact that both parents and professionals were together in one hotel, with a singular focus on a condition that most of my friends had never heard of. It gave me a great feeling to know an organization existed that could make such a meeting happen.

Thirteen years later, as I prepare to begin a term as president of The National Fragile X Foundation Board of Directors, that feeling of being overwhelmed is starting to return! Fortunately, I am joining an experienced board with incredible energy and enthusiasm for the work of the foundation. Steve McKee has provided outstanding leadership over the past two years, and we owe him and his family a great debt of gratitude for all that he has helped us accomplish.

For more than 20 years now, the NFXF has been a trusted source of information and support for families impacted by Fragile X. We have an active public policy initiative designed to increase awareness and enhance funding for FX research and services. In recent years, the foundation has steadily increased its support for innovative research that helps us better understand the various Fragile X conditions and evaluate a range of treatment options.

Though much has been accomplished, we all know that much remains to be done. I look forward to working with parents, the board, and the NFXF staff as we meet the many challenges still before us with the same enthusiasm and commitment that so moved me in Albuquerque 13 years ago. 



The Adolescent and Adult Project

Smoothing the Transition to Successful Adulthood

The National Fragile X Foundation's Adolescent and Adult Project includes extensive video vignettes and written materials designed to assist parents and others in guiding adolescents affected by Fragile X on the path to adulthood. These materials are the result of a more than two-year effort by a team of professionals, parents and NFXF staff members.

For information, log on to: http://www.fragilex.org/html/adolescents_and_adults_project.htm 



Volunteers Make Our World Go 'Round

A little pop quiz to start off the column this issue:

1. When is a human service organization more than the sum of its parts?
2. When is a volunteer more than a volunteer?
3. When is an employee more than an employee?

Don't worry; the answers are easy:

1. When that organization is The National Fragile X Foundation—now combined with Conquer Fragile X Foundation!
2. When that volunteer is Steve McKee, or any other member of the NFXF Board of Directors, one of its committees, parent support groups, Scientific and Clinical Advisory Committee, or is one of many who work on behalf of awareness, support, education, research, legislative advocacy and fundraising.
3. When that employee is Deborah Kwan, former Fragile X programs coordinator at the NFXF.

Please allow me to elaborate.

The National Fragile X Foundation has always worked to unite the Fragile X community, especially through our international conferences. It's part of our mission, so it gets a lot of ongoing attention. But even I didn't foresee the type of uniting that is quickly resulting from Conquer Fragile X becoming an affiliate of the NFXF.

While the NFXF has funded more than \$1 million in clinical and basic science research over the past four years (including the seed grants that recently led to the \$21.8 million NIH award to UC Davis), it still is just one part of our comprehensive mission. At the same time, Conquer Fragile X Foundation has focused almost exclusively on funding basic Fragile X research geared towards improved treatments and an ultimate cure. Now, by working together, we'll be able to support more research than ever before!

I first met Conquer Fragile X Chairman Harris Hollin at our 7th International Fragile X Conference in 2000 in Los Angeles. He was a bustling bundle of energy determined to make things happen with research, especially internationally. Since then, I have watched as Harris did just that: identifying promising research opportunities and supporting that research, from Israel to Belgium to the U.S.A.

Harris was then, is now, and always will remain someone who embodies that great spirit of volunteerism. As such, he joins another incredible group of volunteers at the NFXF.

Ever wonder how we're able to be so many places and do so many things with such a relatively small staff? Simple answer: volunteers.

Outgoing NFXF Board President Steve McKee is a case in point. In addition to being the father of three wonderful children, including a son with fragile X syndrome, and commuting between his office in Washington D.C. and his home in Wisconsin, Steve has somehow managed to lead a process that significantly revamped the way that the NFXF conducts business. Without a solid foundation of good business practices, no nonprofit organization can adequately focus on its human service mission. And while the NFXF's business practices were already good, the rapid expansion of the organization was threatening to overtake our ability to keep track of and manage it all.

Steve McKee to the rescue!

During Steve's almost seven years on the board, and especially during these last two years as president, Steve has led our effort to improve the quality of our database of families and professionals, our financial recordkeeping, and our board governance, including the effectiveness of the

...by working together, we'll be able to support more research than ever before!

continued on following page →

board committees. That's just a few issues that he tackled. Again, this may not seem like such a big thing in the world of Fragile X issues, but anyone in the nonprofit, human service field will tell you that these are the things that, left unattended, can destroy years of good works.

Thanks Steve! Because of your efforts, I, and the entire NFXF organization of families and professionals, can sleep just a bit better each night. (Now if we could just get all the kids with fragile X syndrome to do the same!)

Steve is just one example of the parents and professionals who volunteer on our board of directors (or one of its many committees) and help create the long-term strategy of the NFXF. Other examples include:

- Parents (including boardmembers) who invest so much of their time and energy to organize fundraising events (like our Chicago, San Francisco and North Carolina galas), awareness and educational activities,
- Professionals who volunteer on our Scientific and Clinical Advisory Committee to help guide the organization through the technical and often complicated developments that make up Fragile X research and treatment,
- Advocates who show up in Washington, D.C. each year for Fragile X Advocacy Day (or who participate from afar by regularly writing their congressperson),
- Support volunteers who run our affiliated parent support groups or serve as a parent contact for their region, often organizing Fragile X workshops and seminars such as the recent event in Chicago.

The list could go on, but the message should be clear: The NFXF is able to be a comprehensive organization because volunteers work hand-in-hand with the NFXF staff on behalf of families everywhere.

So who are those staff members with whom the volunteers work hand-in-hand? Well, they're listed on our staff page in this journal—with one notable and new absence: **Deborah Kwan**. As many of you know, Deborah served as the NFXF's fragile X programs coordinator for nearly eight years. When the NFXF relocated from Denver to the San Francisco area in 1999, Deborah was the first person I hired. (Though she first volunteered!)

Over the years, Deborah, in addition to being responsible for countless tasks that are necessary to keep a national organization running, helped coordinate three international conferences, put together the resulting Conference Proceedings, helped birth the “Lesson Planning Guide for

The NFXF is able to be a comprehensive organization because volunteers work hand-in-hand with the NFXF staff on behalf of families everywhere.

Students With Fragile X Syndrome” and the “Adolescent and Adult Project” binder and DVD, and authored and edited numerous pamphlets and articles. Mind you, she did these things in her spare time, meaning, when she wasn't talking on the phone or emailing with the thousands of parents and professionals she has helped these past years.

Deborah: I'll miss you. You helped make the NFXF what it is today, and we couldn't have done it without you. Good luck with all of your new activities!

Elsewhere in this issue, former board president and parent John Harrigan writes from a more personal perspective about what Deborah has meant not only to the Fragile X community, but also to his family.

Readers might be wondering what will now happen to the high-level of telephone and email support that the NFXF has historically provided. Let me reassure you that we have already taken steps to ensure that our support services continue and, in fact, becomes even more comprehensive. (I'll be filling you in on the details in the next issue of the *Foundation Quarterly*.)

Until then, next time you have the opportunity, thank a volunteer! And for much more about our many volunteers, please see page 28, “Focus On Volunteers.” ☞

Robert Miller has been executive director of The National Fragile X Foundation since 1999. Email: RobMiller@FragileX.org



Riding Out the D.C. Political Roller Coaster

Our efforts pertaining to the 2008 federal budget commenced around the first of this year (2007) when we began to craft our specific requests for next year's (2008) budget. Our requests fall into two categories: requests for directive language from Congress to health agencies which are part of the National Institutes of Health, and requests for specifically earmarked funding for health agencies that are part of the Centers for Disease Control. Both significantly impact the amount and focus of federally funded research and programming related to Fragile X.

Working with the many parts of the FX community, consensus was reached on our requests to Congress last January. NFXF advocates presented these requests in March (on NFXF Advocacy Day), and they have been reinforcing them ever since through home state visits, phone calls, letters and emails.

This coordinated effort was quite successful. Nearly all of the directive language we requested to increase FX research and move it in the direction that scientists believe will produce near-term tangible results was adopted. In addition, Congress (both the House and the Senate) recommended doubling the size of the FX public health program at the CDC from over \$900,000 to more than \$1.8 million yearly.

So far, so good....but then politics reared its head. As with so many issues in Washington today, there is much jockeying for position between "R" and "D" as we approach next year's election. And there's certainly no shortage of political tension between Congress and the President. Despite the fact that the proposed budget was a truly bipartisan effort, on November 13, the President made good on his promise to veto the LHHS budget bill that included all of our hard-fought FX provisions. ("Too expensive," he said.) The NFXF Advocacy Team issued an immediate Action Alert, and within 24 hours nearly 2,500 emails were sent by Fragile X Advocates to Congressmembers urging an override. Our letters were

delivered to 82 of 100 Senate offices and 230 of 435 House offices—a remarkable achievement in such a short time. On November 15 at 11 p.m., the House reconvened in an attempt to override the President's veto. ("Critically needed," they said.) House Appropriations Committee Chair David Obey

What's most important is how far we've come and the results that NFXF advocates have achieved in such a short period of time.

(D-WI) set the stage and explained the consequences of failing to override the veto as follows: "For medical research...at the National Institutes of Health, meeting the President halfway would put us \$700 million below the bill we are considering today. That means about 700 fewer grants for research to treat and cure so many deadly diseases."

Despite urging from NFXF Advocates and nearly 1,100 organizations representing tens of millions of Americans, the House came up a few votes short and the President's veto survived. Before Congress recessed for the Thanksgiving holiday, legislators had already begun exploring a compromise with the President. Whether that's possible—and what it will mean to FX—was not yet determined at the time this issue of the *Foundation Quarterly* went to press. Congress was expected to pick up where it left off after returning from the Thanksgiving recess.

All of which compels me to say: *Stay tuned, don't be discouraged, and keep up the great work!* What's most important is how far we've come and the results that NFXF advocates have achieved in such a short period of time. The directive language to NIH that we've been successful in securing since 2004 assures:

- Continued funding of the three FX Research Centers of Excellence,

continued on following page →

- The acceleration of “translational” research leading to clinical trials of possible new drug treatments,
- Better coordination among all agencies conducting FX research.

In addition, this directive language contributed substantially to the largest-ever research grant (\$21.8 million) for Fragile X, awarded recently to the group led by Dr. Paul Hagerman at UC Davis. (See the September issue of the *Foundation Quarterly* for details.) Moreover, the efforts of NFXF advocates are directly responsible for the creation and funding of a National Fragile X Public Health and Research Program at the CDC and now (hopefully) a significant increase in funding for that program. These are remarkable results, and congratulations are due to everyone who has contributed to our many advocacy efforts over the years.

For the most current update on all these efforts, please visit the Public Policy and Legislative Advocacy page at www.FragileX.org. While you're there, you can also register as an advocate to receive regular email Advocacy Updates and Action Alerts as events unfold.

Better yet: Make plans now to join us for the next NFXF Advocacy Day February 26-27, 2008 in Washington, D.C.

There is only one way for Congress to truly understand how important FX is: if we take the time to tell them. And there is nothing like a face-to-face meeting to get your point across. More details on Advocacy Day will follow in the next months, so keep checking the website and your email.

To all our supporters: Truly, you make all that we do possible. Please consider a generous donation to the NFXF Annual Fund, so that together we can continue this most important work on behalf of our entire Fragile X community.

All the best in the new year from your NFXF Washington team. ☎

Jeffrey Cohen chairs the Public Policy Committee of the NFXF. Email: j.cohen@FragileX.org.

Committee members include Serena Lowe, William Parker, Mary Beth Bohm, Michael Kelley, Brad Whitus, John Harrigan and Robert Miller.

About NFXF Advocacy Updates and Action Alerts



Once you've registered as an NFXF Advocate you'll receive regular **Advocacy Updates** and **Action Alerts**. These email communications are the best way to stay informed as events unfold in Washington. When we need you to reach out to your congressional representatives, you'll receive specific instructions and a link to our congressional communication system that will allow you to quickly personalize and send an email message in two minutes or less.

But the system works only if you participate, and most requests to communicate to Congress are time-sensitive. So when you receive an **NFXF Action Alert**, please view the email within 48 hours and then respond by clicking on the link to the congressional communication system, filling out the required information and sending your email.

We have achieved great results with this system, but your continued participation is critical. Don't assume someone else will take care of it—they might be thinking the same thing of you! So please make yourself that “someone else,” and, as the sneaker ad suggests: *Just Do It*.

To register as an NFXF Advocate, log on to: https://secure.cnchost.com/fragile.org/fragile_x_advocate.htm ☎

A Deborah Kwan Appreciation

There are times in the life of any organization when someone has to step up and fully embrace their talent, vision, and dedication—and thus make a difference. We have always been blessed to find these people when we have needed them most—from the inestimable Randi Hagerman to Louise Gane, Gene Koebel and Jerry McCabe in the Denver days, to Jeff Cohen in Michigan, to Robby Miller and Deborah Kwan over the past eight years in the San Francisco area.

Life is full of twists and turns, of opportunities lost and seized, of predictable reactions and positions to totally out-of-the-blue moments of, “What was *that*? It wasn’t long after our sons, Sam and Ben, were diagnosed with fragile X syndrome—one of those “What was *that*?” moments—when we moved over the East Bay hills from our longtime haunts in Berkeley and north Oakland to the ‘burbs. It was 1993.

We settled in pleasantly in Pleasant Hill and went looking for a good pre-school program for Ben after getting Sam situated. We found it at a wonderfully named Sufi-run program called “The White Pony.” Ben’s occupational therapist was Deborah Kwan.

My wife Stephanie and Deb hit it off instantly, as they both shared ballet as a passion. Deb had danced with the Joffrey Ballet in New York and Steph danced locally with, among other characters in our drama, Julie Miller, who had a husband named Robby who was an early intervention special ed guy who later, well...I’ll save that story for another time.

Deborah was terrific with Ben. She worked him hard, and he loved the pressure she exerted, the joint work, all of it. Deb truly *got* it; she understood FXS intuitively, and Ben—along with all her other “patients”—thrive because of it.

Cut to 1999. The National Fragile X Foundation moves to northern California and we hire one Robby Miller as our executive director. The first person who goes to work for him—first as a volunteer, and later as a staffer—is Deborah Kwan. Her surgically repaired shoulder had finally worn out. She couldn’t do OT anymore, but she had a passion for developmentally disabled kids, and she *got* it. All of it. The rest is history—and a treasured part of NFXF history it is.

From organizing three NFXF International Conferences



Sam Harrigan



Ben Harrigan



John Harrigan

(you don’t even want to know how tough that is, but let’s just say she ended up in the hospital with exhaustion after one of them), to her writing and editing many of our special topics pamphlets, Deb tackled projects with a vengeance and drive that is legendary in these parts. I know that what she was most proud of—and what I am most proud of for her—is the incredible job she did on the Adolescent and Adult Project. Brilliant, and moving.

Deb tackled projects with a vengeance and drive that is legendary in these parts.

Deborah, we are really going to miss you around the foundation. So will the thousands of people around the country you talked to every year. Always listening. Always helpful. I know that you wanted to experience just a little more of your son’s senior year in high school, and I applaud you for that. Too many of us work and work and don’t turn around

until they are gone. As always, you and John will continue your *pas de deux* through life with your usual grace and style. We will miss you. You’re the best. ☹

*The author is a past president of the NFXF Board of Directors, and currently heads the Board of Trustees.
Email: jhnfxf@aol.com*

“Support” is one of the key components of The National Fragile X Foundation’s mission. But what does “support” really mean? While the support we provide takes many different forms, a recent exchange with a mother of a child with fragile X syndrome, whose father (the child’s grandfather) was beginning to show signs of FXTAS, illustrates the length members of our organization will go to help families in need.

The woman was associated with the neurology department of a large hospital in the northeast. She was familiar with neurology and could easily converse with her dad’s doctor and with other doctors at the hospital where she worked. That wasn’t the problem. Despite the foundation’s recent mailing of informational postcards to every neurologist in the U.S., her dad’s doctor and her colleagues at the hospital were wholly unaware of FXTAS—and they didn’t seem particularly receptive to learning about it.

And so, to the NFXF she turned. When someone calls the foundation with a question, our staff typically links him or her with someone who can provide a definitive answer. Included in our vast network of resources are experts in virtually every aspect of Fragile X, from behavior to medication, education, molecular biology and much else. In most cases, these experts are but a phone call or email away. In this particular case, that turned out to be true, but with a unique twist we thought readers would enjoy knowing about.

When the woman made inquiries about her father’s neurological symptoms, our staff immediately thought Paul and Randi Hagerman would be the ideal informational resource. It was the Hagermans, after all,

who led their UC Davis team in the discovery of FXTAS. (They actually had the option of naming it “Hagerman syndrome,” but decided that “FX,” rather than their own name, could use the heightened visibility.) But there was only one problem: the Hagermans were on safari in southwestern Uganda (teaching about Fragile X along the way), and were headed into a dense jungle region near Rwanda. No way to reach them by phone. Our staff sent an email inquiry containing the woman’s questions into cyberspace, but with little hope it would be received, they were reviewing alternate contacts.

They needn’t have bothered.

In under 24 hours, the Hagermans’ detailed response

arrived, complete with specific directions to staff back at UC Davis and the M.I.N.D. Institute, indicating which among the several hundred journal articles they’ve authored to send to the woman, along with names and phone numbers of individuals who were available to talk pending their return.

Foundation staff had visions of the Hagermans typing in their response on a laptop from the back of an elephant, but whatever the means, their assistance reached a grateful woman who got her own special glimpse and understanding of what “support” means at the NFXF.

And perhaps best of all for the future dissemination of that other aspect of the foundation’s mission—education—the Hagermans have been

invited to lead a “grand rounds” physician education session at the hospital mentioned above. You can bet that’s another invitation they’ll accept. The logistics are so much easier than an African safari. ☹

—The Editors

**FROM
UGANDA,
With Support**
*Our staff sent an
email inquiry containing
the woman’s questions
into cyberspace,
but with little hope
it would be received...*

The Five Most Common Questions About FXTAS And the Answers...

1 *I am a newly diagnosed adult female carrier. Why should my parents be tested?*

Your father could be a premutation carrier and at risk for FXTAS. If a test confirms that, his physician can monitor him for any neurological or mood changes.

2 *What are the first signs of FXTAS?*

The most common first sign is a tremor while reaching for something. However, in some men the first sign is balance problems, while others show mood instability or irritability, which the family may attribute to “getting older,” “grumpiness,” or short-term memory loss.


3 *How is the diagnosis of FXTAS made?*

The diagnosis is based on three factors: 1) positive carrier testing for the FMR1 premutation, 2) a neurological exam that reveals symptoms of FXTAS such as intention tremor, memory deficit, ataxia, etc., 3) MRI findings consistent with FXTAS, such as specific white matter changes or decreased size of the brain.

4 *One of my male relatives with a premutation is only 45 but has some mild symptoms. Could it be FXTAS?*

There have been reports of men as young as 40 with some mild signs of FXTAS, so it would be prudent for this relative to be monitored closely by his physician.

5 *How do we find out the difference between Parkinson's, age-related dementia and FXTAS in my older male relative?*

First, it is important that anyone at risk for FXTAS undergo FMR1 testing to ascertain whether he is a carrier. (The condition rarely affects females.) The clinical findings for FXTAS are a little different than for Parkinson's, and can typically be distinguished by a neurologist. The MRI findings in FXTAS are quite specific to the condition, and they may help differentiate it from these other adult onset neurological conditions. 


Support the 2007 Annual Fund!

Most every nonprofit foundation is dependent on the people whom it serves to set aside some portion of the profits they enjoy in their own lives and send it on as a contribution that will help sustain the continuing work of the foundation. *And so it is with The National Fragile X Foundation.*

Lacking any golden stream of dollars that rains magically down a mountain into the waiting buckets of NFXF volunteers, the foundation must instead rely on the purposeful and compassionate generosity of its many contributors in the Fragile X community. That is what we are doing again as another year draws to a close.

Our 2007 Annual Fund has been in full swing for several weeks now, with solicitations arriving in the mailboxes of our thousands of supporters throughout the United States and even some foreign countries. Although NFXF fundraising efforts go on all year long in dazzling and imaginative ways (see page 26 for the most recent evidence and inspiration!), the Annual Fund is an absolutely critical effort that helps us in every facet of our mission.

The NFXF would not be the foundation that it is—would not be able to offer as much support, fund as much research, advocate as effectively, provide as much education—were it not for the outpouring of generosity and care our many supporters exhibit every year at this time.

Please support those efforts today! 



ANNUAL FUND 2007

Serving every
sector of the
Fragile X community...





Get Out While the Getting Is Good: How Far to Push the Good Behavior Envelope

Author's note: This column was written in collaboration with my colleague Dr. Karen Riley, assistant professor in the Child, Family and School Psychology Program in the Department of Education at The University of Denver.

It's an observable fact that when children are doing something well, we seem to encourage them to do more. Is it just human nature to expect more even during those times when things are going relatively well? Maybe that is our definition of being a good parent, therapist or teacher. Clearly, when a child is complying and doing what is expected, we tend to accept that behavior as normal, with little notice; but when the child is acting out, we no longer ignore the behavior because it is aberrant, and thus requires our attention. It is the age-old phenomenon: the squeaky wheel gets the grease.

Often, we are asked to explain why a child behaves the way he does. A typical scenario is, "My child seems to be doing well and all of a sudden, out of nowhere, he becomes agitated and upset." As we analyze the situation, it becomes evident that although the child has complied and succeeded at one level, that success is overwhelmed by an expectation that he accomplish even more.

A typical example is when a child seems to transition well to a restaurant, eats politely and interacts during the meal. Upon finishing the meal and leaving the restaurant, you may think, "Since he's doing so well, let's run to the grocery store to pick up some things before we go home." On the way to the store, everything falls apart. Suddenly, you encounter a steadily escalating behavioral nightmare of screaming and hitting that seems unrelated to any specific trigger. The experience prior to going to the grocery store had been pleasant and your child had been enjoying himself. How can things change so dramatically in such a short time?

The answer really lies within the child. He may have succeeded because he understood the expectation and was able to comply long enough to "meet the contract." Or, he may

have been able to abate his sensory needs long enough to get through the meal (with its challenges of background noise, close quarters in a booth or busy conversation around the table). By the end of the meal, the child may have exhausted his self-management abilities and been looking forward to relaxing in a familiar and less demanding environment. When that expectation is eliminated, the child is thrown off balance and becomes unsettled.

Sadly, the next time a "Let's go to the restaurant" plan is presented, the child learns not to trust it, and resists as soon as it's proposed. His non-compliance is likely an attempt to control the circumstance so that it fits within his capacity for self-management.



We know a lot about behavior in children affected by fragile X syndrome. We can now identify antecedents, and we better understand how behavior can be affected by a number of variables that include environment, social interaction, curriculum, sensory factors, language processing, physiological factors and cognitive deficits. Why then, when things are going well, do we forget to bear those factors in mind, only to be forced into considering them as a child's behavior begins to deteriorate? Part of the explanation is our (very human) tendency to be reactive rather than creating and abiding by proactive strategies.

The reauthorization of IDEA (Individuals with Disabilities Education Act) highlights the need to provide positive behavior support. This means that we no longer focus on a reactionary plan, but rather, we “front-load” a good behavioral remedy with proactive strategies to support positive behavior and reduce the likelihood of recurring negative behavior episodes.

There is a fairly simple way to accomplish this. The old saying, “Get out while the getting is good” is our rule of thumb. We want individuals of all ages to experience success and to finish a task or an activity in a positive way. We need to plan ahead and then follow that plan so we don't find ourselves getting “greedy” and expecting even more good behavior from a child than she is capable of producing. The goal is to end an activity or a task *prior* to the child falling apart.

How will we know when to “get out?” (Or: “Exactly how many math problems should we expect her to complete?”) For this information we need to go back to the Functional Behavioral Analysis (FBA). Use the FBA to gather information about when the child is successful and when problems occur.

The question about math problems, although revolving around an academic outcome, is a good example of how to encourage success without going too far. If you are working on math, you want to present the child with problems in a normal context to see how many problems he is able to complete without exhibiting unwanted behaviors such as crying, biting his hand or throwing the manipulatives. At the end of a designated trial period, review the data. What you should look for is the number of problems the child is able to complete without ever exhibiting inappropriate behaviors. That is your starting point for subsequent behavior management strategies during math.

This strategy is a long-term approach to effective behavior management... we need to actually teach a child how to be successful.

For example, the data may show that on three different occasions, the child successfully completed six, four, and ten problems before she started whining or throwing things. In this case, four problems is the starting point. So each time math is presented to the child, the paper should have only

four problems on the page. After the child completes the problems, she should be reinforced by allowing her to stop. The key is that she should stop even if it seems that she could do more on that particular day. The child thus learns that what is presented is what is expected—and no more. The plan is consistent, so she is able to experience completing a task and ending on a positive note. The inappropriate behaviors she has in her repertoire as a defense against overload are not needed, because she has not been pushed beyond her capacity. Moving forward, the number of math problems presented on the page should be increased gradually, with accompanying reinforcers, until the child's performance reflects an appropriate level for her age and development.

If the FBA shows that the child is unable to do even one math problem or to go to the grocery store for even one minute, then we need to go back to understanding why he is unable to complete these tasks and provide him with supports to address the causes of this behavior.

Remember: This strategy is a long-term approach to effective behavior management. Sometimes we need to actually teach a child how to be successful. That means meeting the child where he is: perhaps retreating to and then celebrating the success of just one math problem or one minute in a store if that's what it takes to end on a successful note. This can be discouraging for both parents and professionals, who may feel they are going backwards, but remember, this is one step in a long process. In the end, pulling back and fostering success reduces the opportunities to reinforce inappropriate behavior, which actually allows both the adult and the child to move forward in a proactive way. ☉

The author is a Colorado Springs-based psychologist, special education consultant, and member of the NFXF Scientific and Clinical Advisory Committee.
<http://information@www.marciabraden.com>

When Two...



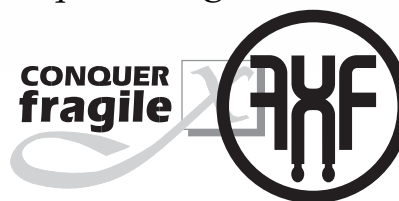
Became One

*Landmark Merger Joins NFXF and
Conquer Fragile X Foundation*

October 31, 2007 was a good day for the world of Fragile X. That's the day two of the world's leading organizations in the field of Fragile X support, research, advocacy and awareness signed a historic merger agreement. The National Fragile X Foundation (NFXF), based in Walnut Creek, California, and the Conquer Fragile X Foundation (CFXF), based in Palm Beach, Florida, signed papers that officially joined them together as one entity, albeit with unique provisions that promise to make the expanded organization much greater than the sum of its parts.

The agreement calls for the NFXF to add CFXF as an affiliate fundraising and research arm. Those activities have been a powerful focus for CFXF since its founding by Chairman Harris Hollin in 1999. Hollin, a former pharmaceutical company executive, came to Fragile X after his grandson was diagnosed with fragile X syndrome (FXS) earlier in the decade. He has since been a tireless advocate and fundraiser for scientific research.

Working closely with Scripps Florida biomedical research institute, the CFXF sponsors an annual Fragile X Young Researchers Conference in Palm Beach, among many other activities. Those efforts are expected to continue and even gain momentum with the merger, given the NFXF's own research funding and its activities through nearly 50 state chapters. The biennial NFXF International Conference should also serve as a powerful complement to the CFXF Research Conference, increasing both fundraising resources and scientific collaboration in the effort to add to the growing



storehouse of clinical and research knowledge about the three Fragile X conditions: FXS, fragile X-associated tremor/ataxia syndrome (FXTAS), and fragile X-associated primary ovarian insufficiency (FXPOI).

The NFXF was the world's first Fragile X organization. It was founded in Denver in 1984, largely as a source of education and support for parents and others, such as health care professionals, who needed assistance with children or patients with fragile X syndrome. Since then, its mission has greatly expanded to include an abundance of awareness activities, legislative advocacy, and, in the past four years, research funding.

Negotiations on the merger took place over many months, with NFXF Board President Steve McKee and Boardmember Jeffrey Cohen taking highly active roles in discussions with Hollin. Hundreds of emails and phone calls and periodic sit-down meetings slowly ironed out the abundant practical and legal issues that always surface when two organizations decide it is in the best interests of their constituents to join forces and become one.


"There comes a point when you want to be more focused than ever on your primary goals, and then you look for some way to achieve that focus," said Hollin, who became a director emeritus of the NFXF with the merger.

CFXF President Karen Fay will become "Conquer Fragile X Programs Director" within the NFXF.

"Raising funds to support scientific research has always been the bedrock of the Conquer Fragile X Foundation, and it became obvious to me in recent years that we could be even more effective if we could reduce the administrative demands on our organization by affiliating with a larger partner," Hollin continued. "The NFXF has a solid infrastructure with a truly national scope. Their resources will allow CFXF not only to focus more on fundraising, but also to offer more support and education activities for the many parents who have always supported us. I'm personally looking forward to increasing my time in fundraising efforts that will continue to expand our scientific knowledge of Fragile X."

NFXF Executive Director Robert Miller was just as enthusiastic as Hollin about the strengthened organization that is already beginning to emerge. "We've always admired the Conquer Fragile X Foundation as a valuable ally in helping to advance everyone's knowledge of Fragile X," he said. "When the opportunity presented itself to formalize a relationship, our board of directors saw nothing but positives for all concerned. Our research side is strengthened, overhead costs are reduced, their parent support and education programs are enhanced, and the entire Fragile X community benefits from a more unified and cohesive approach to the many challenges that Fragile X conditions continue to present."

In a followup note to his board, Miller wrote, "I would just like to thank each of you for helping create the type of organization that others would want to be part of. Harris has been effusive in his praise of the NFXF and Karen has told me how excited she is to be part of the NFXF. I think that says much...Now on to making sure that all of this work brings a tangible benefit to the Fragile X community of families and professionals!"

Under terms of the agreement, both organizations' boards of directors will eventually merge into a single entity under the banner of the NFXF. Hollin will continue to operate from his twin bases in Palm Beach and Philadelphia as he concentrates on strengthening his Fragile X fundraising network along the eastern seaboard. The research funding that emerges from those efforts should go even farther now as the formerly two organizations mesh their fundraising and scientific research decision-making. 

Quotes on a Merger

A sampling of the written reaction that came over the NFXF email network in the wake of the NFXF-CFXF merger news:

"I can't believe this! This is such an important merger that will be of direct benefit to families, clinicians and researchers. Thanks for all your hard work."

—Stephanie L. Sherman, PhD

Department of Human Genetics

Emory University, Atlanta

"For all he did for his grandson, Harris Hollin deserves to be named "Best grandfather of all time." The rest of the FX community has benefited from his hard work as well, and I'm excited that all of that positive energy and the significant dollars raised by CFXF will now greatly increase the NFXF's ability to fund research and provide services to families."

—Jeffrey Cohen

Past President, Current Boardmember

The National Fragile X Foundation

"Congratulations to both organizations; it is satisfying to see highly motivated people able to negotiate towards a common goal. It is additionally rewarding for all of us to see this and to know that now even more people can work together for greater benefit of the FX community. Well done to all involved."

—Dr. Jonathan Cohen

Medical Director, Fragile X Alliance Clinic

Victoria, Australia

"Congratulations to both of you. This is great news. Best wishes for all the success in the world."

—George Jesion, PhD

The Association of University Centers on

Disabilities in Washington, D.C.



Brain Imaging Studies Open New Frontier in Fragile X Research

Research on developmental disabilities, including fragile X syndrome, has been transformed by technological advances in recent years. We can now look “inside” genes to understand their properties. We can examine the biochemical changes that occur as neurons “talk” to each other in response to stimulation from the external environment. We can even create conditions under which we can watch as neural cells are “born.”

Particularly important among these innovations are brain imaging techniques, such as functional magnetic resonance imaging (fMRI), positron emission tomography (PET), single photon emission computed tomography (SPECT), and magnetoencephalography (MEG). These techniques allow us to literally watch the brain as it works to solve important everyday problems, such as recognizing faces, perceiving emotions, calculating a sum, and understanding a sentence. They are also important tools for understanding fragile X syndrome.

These different brain imaging techniques all provide information about where and when activity is occurring in the brain. By controlling what the person being imaged is doing, we can learn which structures or systems in the brain are responsible for solving particular problems or tasks. For example, by comparing patterns of brain activation when a person is viewing pictures of human faces relative to pictures of inanimate objects, we can learn whether there are specialized systems in the brain for dealing with social as opposed to nonsocial information.

The imaging techniques differ in the types of brain activity they detect, how precisely they locate the activity within the brain (spatial resolution), and how reliably they link the activity in the brain to real-time changes in the experience of the person being imaged (temporal resolution).

The techniques also differ in the requirements they impose on the person being imaged. So, for example, fMRI requires that the person being imaged lay perfectly still while completely enclosed in the MRI—something that is difficult for many people with fragile X syndrome, although training and desensitization can help. Despite the constraints, these techniques have been used successfully with people who have

fragile X syndrome, telling us much about how their brains may work differently to solve everyday problems.

The National Fragile X Foundation recently funded a study using fMRI. It was conducted by Laura Holsen, PhD, who was a postdoctoral fellow at the University of Wisconsin-Madison’s Waisman Center (where I work) when she did her research and is now at the Harvard Medical School. Dr. Holsen was interested in understanding more about the difficulties that people with fragile X syndrome have in social situations. She showed people with FXS—mainly

As with virtually any research tool, brain imaging techniques have their advantages and disadvantages, their strengths and limitations...it is important that we be appropriately cautious in interpreting the results of such studies.

teenagers and young adults—pictures of faces while their brains were being imaged using fMRI. She also later tested their memory for the faces. She compared the patterns of brain activation observed in the participants with fragile X syndrome to the patterns of activation observed in typically developing teens and young adults. (See her review of her findings on page 16.)

Why is research such as Dr. Holsen's important? What can we do with information about how the brain works in fragile X syndrome?

I can think of two implications of such research. First, as we begin to test new treatments for FXS, we can make decisions about their effectiveness, at least in part, by determining whether they change patterns of brain activation, making those patterns more typical. Based on Dr. Holsen's research, for example, we might be interested in whether a drug proposed as a treatment for FXS increases activity in the ventrolateral prefrontal cortex while learning in social situations.

Second, research like Dr. Holsen's can help us begin to think about approaches to treatment, such as drugs that specifically target activity in the ventrolateral prefrontal cortex. Of course, there are many steps and much research needed to move from a single study of brain activation to a treatment, but it is a beginning.

As with virtually any research tool, brain imaging techniques have their advantages and disadvantages, their strengths and limitations. It is important, therefore, that we

be appropriately cautious in interpreting the results of such studies. Finding an unusual or different pattern of brain activation in people with FXS, for example, does not mean that the problem is somehow beyond treating, or even that the problem can only be treated with drugs or other biological interventions. Educational and behavioral interventions might also prove to be helpful, even in the case of a brain-based problem.

Brain imaging techniques are bringing us closer to our goal of understanding how the mutation in the FMR1 gene leads to the problems in learning and behavior that are the hallmarks of fragile X syndrome. These techniques are also likely to be part of the answer in finding effective treatments. We at the foundation are pleased that we were able to support Dr. Holsen's study.

The author is a professor in the Department of Educational Psychology at the University of Wisconsin-Madison, an associate director of the university's Waisman Center, and co-chair of the NFXF Scientific and Clinical Advisory Committee. Email: Abbeduto@waisman.wisc.edu

WHAT YOU CAN FIND AT www.FragileX.org



CONFERENCES & WEBCASTS

Find the latest information about The National Fragile X Foundation's upcoming 11th International Fragile X Conference, as well as information about our regional conferences and seminars, as part of the submenu of "Conferences & Webcasts." Here you'll also find the "Proceedings" of past conferences, information about events around the world, announcements about webcasts, and recordings of recent webcasts.

- **NFXF International Conference**
- **Prior NFXF International Conferences**
Previous Conference Photos, 2006 Proceedings, 2004 Proceedings, History
- **Other Conferences and NFXF Exhibits**
- **Around the World Calendar of Fragile X Events**
- **Webcasts**
Upcoming Webcast, Prior Webcasts

RESEARCH

Editor's Note: Dr. Holsen's report below represents an NFXF-sponsored research project nearing completion. Readers of this journal will be alerted when it appears in its final version in a peer-reviewed academic journal.

Interaction Between Emotion and Memory in Fragile X Syndrome: An fMRI Study

**LAURA M. HOLSEN, PhD, Departments of Psychiatry and Medicine,
Connors Center for Women's Health and Gender Biology, Brigham and
Women's Hospital, Boston, MA**



Many individuals with fragile X syndrome (FXS) exhibit social anxiety, which parents and individuals with FXS report as one of the most distressing issues they encounter. Our research focused on whether such social anxiety behaviors are related to difficulty in

remembering faces, given evidence that individuals with FXS exhibit some memory-related deficits. We wanted to know what happens in the brains of individuals with FXS when they are asked to perform a memory task focused on human faces.

We divided our study participants into two groups: one group with FXS and a control group without the condition. Groups had similar numbers of males and females, and were equivalent in age (average age: 19). Using functional magnetic resonance imaging (fMRI), we scanned the brains of individuals in both groups while we showed them pictures of human faces. The scanner monitored what parts of their brains were active while they looked at the faces, and also monitored what parts of the faces they looked at. Then, once they exited the fMRI scanner, we showed them the pictures of the faces again and asked them to tell us if they recognized specific faces that they saw while in the scanner. We were interested not only in how the group with FXS would perform on a face memory task, but also whether they looked at parts of the face (such as the eyes) to a greater or lesser extent than the control group, whether certain brain regions reacted differently when subjects with FXS viewed faces that they remembered later (compared to faces that they did not remember later), and whether these brain responses were related to an individual's level of social anxiety.

SYNOPSIS

This study set out to examine whether the social anxiety and memory deficits characteristic of fragile X syndrome may have a basis in the nervous system. With the assistance of functional magnetic resonance imaging (fMRI) technology, it found differences between individuals with FXS and a control group in both brain activity and memory for faces.

Our study indicated two differences at the behavioral level between the group with FXS and the control group: 1) they had a harder time remembering faces, and 2) they spent less time looking at the eye region of the faces we showed them.

Two additional findings showed both similarities and differences between the two groups in brain processing. In response to faces that they remembered, both groups exhibited similar levels of activity in the hippocampus, the brain region most involved in long-term memory processing. This suggests that in individuals with FXS, the hippocampus responds in a typical manner when forming memories for faces.

However, we also found that compared with the control group, the group with FXS showed some different activity patterns in other areas of the brain. In particular, the ventrolateral prefrontal cortex, a brain region involved in both complex processing during social interaction and working memory, showed less activity in the group with FXS, especially in those with high levels of social anxiety. We think this means that for people with FXS, increased anxiety in social situations may be related to a disruption in brain functioning during the earliest parts of forming memories of faces, especially in parts of the brain that are important for complex social skills.

Functional MRI studies have increased our knowledge about the relationships between genes, brain, and behavior in individuals with FXS. These studies demonstrate which areas of the brain in FXS demonstrate different activation patterns that are linked to specific behaviors relevant to the condition. While tentative, it is likely that these regions of the brain are the most affected by the reduction in the protein of the fragile

continued on bottom of opposite page →

■ NFXF-Sponsored Student Research Report—Two Years Later



By Julie Hanck ■ McGill University Montreal, Canada

WILLIAM & ENID ROSEN SUMMER STUDENT FELLOWSHIP

Editor's note: Members of the NFXF staff were pleased to see a former recipient of a summer student fellowship as a featured speaker at a recent international meeting of clinical researchers in Lake Tahoe. As is the hope and the plan when we provide these fellowships, Julie Hanck has stayed actively engaged in Fragile X research! The report below recaps her continuing investigation into the topic of her summer research. We look forward to hearing more about her accomplishments in the field of Fragile X for many years to come.


The National Fragile X Foundation granted me a Rosen Summer Student Fellowship in 2005. With this wonderful opportunity, I was able to begin my work as a master's student at McGill University (Montreal, Canada). Dr. Kim Cornish was my supervisor at McGill's Child Laboratory for Research and Education in Developmental Disorders.

The first step in completing my research was to create a database of potential participants. Quebec is a bilingual province, with a distinct separation between the French and English communities. Our first goal was to bring both communities together for the purpose of conducting Fragile X research that addressed their common needs. Dr. Cornish and her lab members organized two conferences at McGill in the hope of doing just that. These bilingual events brought together families, teachers and educators from both communities. It has been our pleasure since then to have become acquainted with a number of wonderful families.

My research project is focused on understanding the development of two cognitive domains known to be affected in fragile X syndrome: attention and visual perception. We are interested in improving our knowledge of the way children with FXS see and understand the world around them. Our research focus is to chart the development of their attention and visual abilities in early development all the way through to age 15.

We have learned a lot about children with FXS over the past decade. We believe that in organizing visual information, they use their brain differently than typical children do. We know that one particular strength is their ability to recognize objects. However, by the time children with FXS reach age 10, they often demonstrate difficulty in catching objects (e.g. a bouncing ball).


Preliminary results suggest that just as with the larger population, performance on attention and visual perception tasks for children with FXS is not static, but is dynamic throughout development. This is encouraging, because it highlights the importance of not only studying FXS at single time points such as in infancy or adulthood, but also, to study progression of various skills through time. Tracing the development of these skills and challenges in attention and visual attention will help us to fully understand the cognitive profiles of FXS and to develop specific interventions that can be implemented to maximize every child's potential.

Through this research we look to help inform and educate families, teachers and clinicians about the importance of interventions that target the unique needs of children with FXS beginning as early as possible in their development, and then continuing with a developmentally appropriate program into adulthood. 

RESEARCH

continued from previous page

X mental retardation gene (FMRP) that characterizes FXS. Although there remains a great deal of work to be done in delineating how this disruption evolves over time, we believe this study provides evidence that social anxiety in FXS has a

neural basis. Our hope is that with further research, we may better understand the basis of social anxiety in individuals with FXS, thus leading to more effective ways to address difficulties they experience in social situations. 

■ Special Topics Pamphlet

Editor's note: The article below is derived from the latest addition to The National Fragile X Foundation's "Special Topics" Pamphlet Series. Other topics in the series are listed on the resources order form on page 31.

As always in the special topics series, this pamphlet is intended to be an introduction and overview to a complex topic. Therefore, each pamphlet includes a listing of additional resources. It is our hope that parents use these pamphlets as guideposts and references as they learn how to assist those (including themselves) who are impacted by Fragile X. All special topics pamphlets are available from The National Fragile X Foundation free of charge.



Fragile X Testing: Facts for Patients and Health Care Providers

Many physicians are now offering their patients a test called the "Fragile X test" or "FMR1 testing." This booklet will explain what the Fragile X test is and what it will tell you about your or your child's genetic background. It is done in a basic question-and-answer format.

WHAT IS FRAGILE X?

Fragile X is a family of conditions that includes:

- **Fragile X syndrome (FXS):** the most common inherited form of mental impairment. FXS occurs in both genders, though boys tend to have more severe symptoms. It causes behavioral, developmental and language disabilities.
- **Fragile X-associated primary ovarian insufficiency (FXPOI):** a cause of infertility, early menopause and other ovarian problems in women of reproductive age.
- **Fragile X-associated tremor ataxia syndrome (FXTAS):** an adult onset (over 50 yrs.) neurological condition causing balance problems and tremors.

These three conditions are all caused by changes in the fragile X (FMR1) gene. The FMR1 gene is on the X chromosome.

WHAT KINDS OF CHANGES OCCUR IN THE FMR1 GENE?

The FMR1 gene has a pattern of DNA, called a CGG repeat. Each person has a different number of CGG repeats. Some individuals have an expanded number of repeats that is above the typical range.

The FMR1 gene exists in four forms. These are defined by the number of CGG repeats:

1. Normal range

- This gene has 5-45 CGG repeats.

2. Premutation

- A premutation gene has 55-200 CGG repeats.
- Individuals with an FMR1 premutation are also known as FMR1 "carriers."
- Approximately 1/250 women and 1/800 men in the general population are premutation carriers.
- It has the potential to be unstable and to expand when passed from mother to child, thus causing fragile X syndrome.
- Many individuals with a premutation have no symptoms and no known family history of Fragile X.
- Females with a premutation are at risk to have children with fragile X syndrome and are also at risk for infertility and other ovarian disorders.
- Males with a premutation are at risk for an adult onset neurological disorder called FXTAS. Also: they will pass the premutation on to all their daughters and none of their sons.

3. Full mutation

- A full mutation gene has more than 200 CGG repeats and leads to the symptoms, such as mental retardation, that are associated with fragile X syndrome.
- Approximately 1/3600 males and females have a full mutation.
- A full mutation will cause fragile X syndrome in males.

- Females with a full mutation can exhibit a wide range of intellectual, physical, behavioral or social effects of fragile X syndrome. Approximately 50 percent of females with a full mutation will have IQs in the mentally impaired range.

4. *Intermediate or "grey zone"*

- This gene has 45-54 CGG repeats.
- Approximately 1/50 individuals has an intermediate sized gene.
- This gene is not associated with any known medical problems, and individuals with it are not at known risk to have children with fragile X syndrome.
- In a small number of families, this gene changes slightly in the next generation and may have the potential to lead to premutations in future generations.

WHO IS AT RISK TO BE A FRAGILE X CARRIER?

Anyone (1/250 women, 1/800 men) can be a Fragile X carrier. Among those at increased risk are:

- Individuals with a family history of Fragile X or mental impairment, including autism and other developmental/behavioral disabilities.
- Women with infertility or FXPOI and men with neurological findings consistent with FXTAS.

HOW IS FRAGILE X SYNDROME INHERITED?

Though both males and females can be FMR1 carriers, fragile X syndrome is passed on from a mother to her children. A mother can have either a premutation or a full mutation and pass it on to her sons or daughters.

WHO SHOULD TAKE THE FMR1 TEST?

You may wish to take an FMR1 test if:

- You are planning, or early in, a pregnancy.
- You have a family history of mental impairment, autism or learning disabilities.
- You are having difficulties conceiving, or have been told you have ovarian dysfunction.
- You are a male over 50 and have neurological signs of FXTAS.

WHAT DO TEST RESULTS MEAN?


- If you have results in the normal range, you are not at risk to have a child with FXS.

- If you have a "grey zone" result you are not at risk to have a child with FXS (and therefore no prenatal testing is warranted). However, future generations of your family may be FMR1 carriers.
- If you have a premutation and you are male, then all of your daughters will be carriers and none of your sons will inherit your FMR1 gene (since they receive your Y chromosome). Also, you may be at risk to develop some of the neurological signs of FXTAS.
- If you have a premutation and are female, you are at risk to have a child with FXS, and are also at risk for infertility and early menopause.
- If you have a full mutation and you are female, you may have physical or learning issues related to FXS. You are also at risk to have a child with FXS.
- If your child has been diagnosed with a full mutation, he or she may likely exhibit some level of mental impairment, language or behavioral issues, and will thus benefit from professional services.

WHAT ARE THE REPRODUCTIVE OPTIONS FOR FEMALE FMR1 CARRIERS?

- You can attempt to conceive a pregnancy and choose no intervention.
- You can have prenatal diagnosis (amniocentesis at 15-20 weeks or CVS at 9-11 weeks) in a pregnancy.
- You can elect to use a non-carrier egg donor.
- You can pursue preimplantation genetic diagnosis (PGD). This assisted reproductive technology is available at a limited number of centers nationally. It involves withdrawing eggs from the carrier and then after fertilizing and testing the fertilized egg, implanting only unaffected eggs.
- You can choose to adopt children.
- You can choose to not have any or additional children.

HOW CAN A PERSON LEARN MORE ABOUT FRAGILE X?

Anyone considering or undergoing carrier testing, or who has a positive FMR1 test, may benefit from meeting with a genetic counselor. Your physician or health care provider can refer you to a genetic counselor in your area or you can find one at www.NSGC.org. You can also call the genetic specialist at the NFXF at (800) 688-8765. 



A Primer on *Sensory Integration*

Sensory integration is the ability to organize light, sound, movement, and texture unconsciously and automatically, with no obvious effort. This organization is accompanied by age-appropriate behavior and emotional responses, social engagement, and cognitive reasoning skills, all of which should lead to purposeful work and play.

Without sensory integration, children may experience:

- A need for constant movement or sedentary withdrawal,
- Speech delays that may be accompanied by social avoidance or awkwardness,
- Clumsiness and uncoordination,
- Decreased attention to task or no visual attention at all,
- Poor safety awareness,
- An elevated tolerance to pain.

More often than not, we have children walking through the doors of our therapy center with diagnoses such as fragile X syndrome, ADD/ADHD, Down syndrome, autism spectrum disorders, muscular dystrophy, cerebral palsy, bi-polar disorder, fetal alcohol syndrome, reactive attachment disorder, seizure disorder, anxiety disorder, and many more (including no diagnosis at all). Given the dynamics of each disorder and inherent differences in personality, every one of those children is different, with specific needs we must tailor to their individual situation.

Could all of these children, with their varied diagnoses, share an additional diagnosis of sensory integration dysfunction (a.k.a. sensory processing disorder)? Absolutely.

For one reason or another, many of these children have missed or skipped critical milestones in their development. The ramifications of this can be profound, given our knowledge that one integrative task typically builds upon and interacts with others, allowing the child to build a repertoire leading to sensory mastery. Failure to achieve key components in that mastery is like building a house with a flawed foundation. The result can lead to sensory integration disorder.

Many children take medications and/or go on special diets to curb the associated behaviors related to sensory disorders. Both of these approaches may be beneficial. Working in close consultation with a physician and therapist, parents can determine the possible efficacy of any approach by trial and error.

All of us have unexplainable responses to certain stimuli. The near-universal revulsion to fingernails scraping a chalkboard is a classic example. Others are personalized and even quirky, such as our abhorrence of certain smells or textures of foods that most other people enjoy. Some people, mostly men it seems, stick their tongues out during deep-in-thought activities such as cooking or writing. When we are busy, stressed or bored, some of us chew gum until it becomes the texture of rubber.

There is a qualitative difference between behavioral quirks that reflect an individual's unique sensory processing mode and a full-fledged sensory integration disorder.

Without the automatic ability to tolerate the environment (whatever and wherever it is), the effort it takes to learn reading and writing is spent instead on avoiding lights, blocking out sounds, or holding one's body still enough to sit in a chair.

On an almost daily basis, I hear these kinds of anguished comments from parents:

"Why didn't anyone tell me about this two, five, 15 years ago?"

"I know there is something wrong, but no one can put their finger on what is happening with my child or what to do for him."

"As a mother, I know something isn't right, but I have been told not to worry, that my child will grow out of it. But she hasn't."

A person knowledgeable in sensory integration can give you a sensory-based explanation for all these behaviors. For example, we may swing our leg or tap our foot under the table at a meeting to help regulate our sensory system. This feeds a proprioceptive need (feedback to muscles and joints) and helps keep us awake, interested and organized in thought. Or we may feel the need to chew on gum or suck on candy during a seminar. The mouth is the organization center, keeping our system busy, our mind alert and organized.

So, if you can identify with the quirky behaviors mentioned above, or have 10 more of your own, do you have sensory integration disorder? Not necessarily.

There is a qualitative difference between behavioral quirks that reflect an individual's unique sensory processing mode and a full-fledged sensory integration disorder. The latter manifests in extreme responses that affect one's ability to participate and be effective in daily life. Examples would be a child incessantly walking the perimeter of a playground instead of playing on it with peers, and severe food pickiness that leaves a child eating only a few foods.

Often, people with SID will avoid and seek. They may avoid saying hello or interacting with visitors, but love to crash and ping from one couch to the other while a visitor is there. Or they may avoid certain textures in food, but over-stuff their mouths with other foods.

Scientifically, we can prove a chromosomal or genetically imposed diagnosis, but it is more difficult to explain the behavioral characteristics that come with those diagnoses. Behavior, after all, results from an extremely complex interplay of nature, nurture and culture.

Sensory integration treatment will not remove a diagnosis, but through treatment and education, it can decrease inappropriate behaviors. It is our job as sensory integration specialists to teach children how to process information, how to identify and respond to various forms of sensory input as needed to make it through life one minute at a time. Our hope is to empower them with the tools they need for independence and stability.

Living with a person who has special needs can be challenging, so parents are always looking for additional tools to help make their children's lives (and their own) more efficient and purposeful. Every day, all of us change... learn... adapt. It is imperative that just as we do with eating and



breathing, we integrate our sensory lives so that the basics of living are just that: *basic*. All of our sensory systems were put in place for a reason, and without them we couldn't ever make sense of what we feel, see, hear, taste or smell. All these functions must be in sync for our "selves" to work, play and just *be*.

Sensory integration is essential for growth. It involves much more than a brush on the skin or hands in a sandbox. It is very much a neurological concept, and an integrated approach to how we learn about and function in our world. (FF)

The author holds a master's degree in occupational therapy and is certified to administer the Sensory Integration and Praxis Tests (SIPT). She owns Interactiv Children's Therapy Services, Inc. with two locations in the Atlanta area (www.ictsonline.com). Email: tmulvany@ictsonline.com



From POF to POI: Evolution of a Term

About 10 years ago we began seeing the first reports of increased rates of infertility and early menopause in women with FMR1 gene premutations. Since then, we have learned that approximately 22 percent of premutation carriers have some degree of ovarian dysfunction. Many of these carriers were diagnosed with “POF” (premature ovarian failure), meaning their ovaries were functioning similarly to those of a woman in menopause. However, over the last few years, specialists in the field have become increasingly uncomfortable with referring to the ovarian complications in FMR1 carriers as “POF.” There are several reasons for this evolution in their thinking.

First, the term “POF” does not include the full spectrum of clinical ovarian symptoms in women with premutations. Some carriers merely have reduced fertility or irregular cycles, which are milder symptoms than complete premature ovarian failure. Additionally, the word “failure” suggests that it is impossible for women to get pregnant, but that has turned out not to be true. About 5 percent of women with “POF” do manage to conceive a child after receiving this diagnosis.


For these reasons, reproductive specialists have decided that *a more accurate term for this condition is primary ovarian insufficiency, or “POI.”*

“Primary” means that the disorder originates in the ovary, as opposed to some other organ or hormonal source, such as the brain.

“Insufficiency” means that the ovaries are not functioning to their normal capacity. This differs from “failure,” which indicates a complete shutting down of the ovary.

Recommendations to replace the term “POF” with “POI” have recently been published in infertility medical journals. This trend is occurring outside the field of Fragile X, among scientists and clinicians working with populations of women who have POI for reasons other than an FX premutation.

Eventually, POF may be completely replaced by POI in the entire medical community and the patient information materials that flow from its research. That is why, after careful deliberation and much discussion with those most knowledgeable in the field, we have decided to use this more scientifically accurate term in all of our own materials at The National Fragile X Foundation. In the coming months, readers will see the change reflected in this journal, our website, and in the educational literature we produce for families. And since the NFXF will be addressing POI only as a manifestation of Fragile X, it will be referred to as “fragile X-associated primary ovarian insufficiency” (FXPOI).


We trust that this change is easily understandable, and reflective of the ever-evolving knowledge we are gaining almost daily in this relatively new and dynamic field of Fragile X studies. 

The author is a certified genetics counselor and the staff genetic specialist at the NFXF. Email: liane@FragileX.org

Changed Your Email Address?

Attention Foundation Members! Have you recently left your former email home for some greener pasture in cyberspace? Have you perhaps notified all your personal correspondents, but left your institutional friends off the list? We won't be hurt, but please do update your address with us today so you don't miss out on NFXF Membership E-benefits that include:

- Courtesy email reminders of upcoming foundation events and activities,
- Our quarterly email newsletter *Fragile Xtras*, featuring details on the latest happenings.

It's easy to stay informed: Just send an email to Membership@FragileX.org and reference NEW EMAIL in the subject line. You don't even have to write a note—though we are ALWAYS happy to hear from our members, so keep on typing if you are so inspired! 

The National Fragile X Foundation Presents the
11th International Fragile X Conference

Hosted by The Fragile X Resource Center of Missouri

St. Louis, Missouri ■ July 23–27, 2008

REGISTRATION INFORMATION

Online registration for the 11th International Fragile X Conference will be available January 31, 2008 at www.FragileX.org > **11th International Fragile X Conference**. Registration forms will also be available upon request.

HOTEL INFORMATION

The conference will be held at the **Hyatt Regency St. Louis at Union Station**. Rooms are available for conference attendees at the special rate of \$125 per night, plus taxes. To make a reservation, please see the contact information below. Be sure to let the hotel representative know that you are with **The National Fragile X Foundation Conference** to get the special room rate!

Hyatt Regency St. Louis

One St. Louis Union Station

St. Louis, MO 63103

(800) 233-1234 or (314) 231-1234

www.Hyatt.com

SCHOLARSHIPS

A limited number of scholarships are available to assist with conference registration fees from the NFXF. Applications for these scholarships will be available beginning January 1, 2008 at

www.FragileX.org > **11th International Fragile X Conference**. Applications are due no later than March 31, 2008. Priority will be given to first-time applicants. Scholarship recipients are expected to volunteer for a minimum of 8 hours at the conference.

CHILD CARE

Child care will be available at the Hyatt Hotel for children ages 2–16 years. Registration for child care will be available on a first-come, first-serve basis on January 31, 2008 at www.FragileX.org > **11th International Fragile X Conference**.

TRANSLATION

Spanish translation will be available for selected conference sessions and workshops. Specific sessions will be determined at a meeting on the first day of the conference, by the consensus of those requesting translation. Please make certain that you request translation when completing your registration form.

EXHIBITOR SPACE

Exhibitor space is currently available. For information, including booth size and rates, call the NFXF office (925) 938-9300 or email NATLFX@FragileX.org.



11TH INTERNATIONAL
FRAGILE X CONFERENCE

July 23–27, 2008  The National **Fragile X** Foundation

Getting to St. Louis Without Breaking the Bank

Before the holidays (and their bills) start to arrive, we thought that we would provide information on ways to make it financially feasible to attend the **11th International Fragile X Conference** in St. Louis, MO next July.

THE IRS

Let's take a look at some available tax deductions:

Section 213(d)(1)(B) of the IRS code, provides that "medical care" also includes transportation that is "primarily for and essential to" its provision.

Section 213 (d)(2) provides that the cost of lodging (up to \$50 per night) while away from home, that is primarily for and essential to medical care, is tax-deductible if: (A) the care is provided by a physician in a licensed hospital or a related or equivalent facility, and (B) there is no significant element of personal pleasure, recreation, or vacation in the travel away from home.

What this means: Let's say your son, Josh, has been diagnosed with fragile X syndrome. You, on the recommendation of Josh's doctor, travel to St. Louis to attend the conference, where you will obtain medical information that may be useful in making decisions concerning Josh's treatment or care. While there, your social and recreational activities are secondary to your attendance at the conference. Given these facts, the registration fee paid to attend the conference is primarily for medical care, and your travel is primarily for, and essential to, medical care. Accordingly, you may deduct the registration fee and transportation expenses under section 213 (subject to the limitations of that section).

Unfortunately, meal expenses while receiving medical care are not deductible unless they are provided at a hospital or similar institution at which the taxpayer, taxpayer's spouse, or dependent is receiving the care.

BEYOND THE IRS

What if tax deductions aren't enough? What if you require additional funds in order to attend?

Many of our members have found success by contacting various service organizations to request monetary assistance. Possible resources include: The Knights of Columbus, The Lions Club, The Rotary Club, Shriners, Elks, Moose, and the

VFW. Other sources may be a local developmental disability group, a county health program, or a private medical or therapy company. Now is the time to ask—it is not too early!


However, one shouldn't expect to obtain the total funds required for the trip from a single source. It thus follows that contacting several agencies may increase the chances of receiving adequate funds. (*Note:* You may need to pay for many, if not all, expenses up front and then seek reimbursement.)

Finally, if your child has an early interventionist, county social worker, or case manager, ask them if they are aware of any other public monies that may be available for parents of children with disabilities. Many states have funds available.

The following lists key points for a letter you could adapt for your personal use.

- Reason for writing ("...to request funding to attend the 11th International Fragile X Conference sponsored by The National Fragile X Foundation (NFXF)").
- Your child's condition, year of diagnosis, and a general description of learning and behavioral problems associated with FXS.
- Date & location (July 23-27, 2008, St. Louis, MO) and general conference info ("brings together parents, teachers, speech pathologists, occupational therapists, physicians, and researchers to address all aspects of fragile X syndrome").
- Anticipated expenses (registration, lodging, food, travel)
- Reason attendance is important (gleaning information from professionals, networking/learning from other parents).
- Reiteration of request for financial assistance and note of thanks.

Finally, you may wish to offer to do a presentation for their group following your trip (if the organization desires). It could be oral or written. *Note:* The NFXF has a PowerPoint presentation (or slides) available for such occasions.

The National Fragile X Foundation hopes that this article helps make attendance possible for readers who may otherwise have difficulty financing a trip to St. Louis. 

Former NFXF staff member Deborah Kwan helped coordinate the last three International Conferences.

Call for Abstracts

THE NATIONAL FRAGILE X FOUNDATION

St. Louis, Missouri, USA • July 23-27, 2008

The 11th International Fragile X Conference is designed to provide a forum in which members, colleagues, and associates of the foundation can present their scientific, clinical or scholarly work; to present a general program that will be informative and of interest to all conference participants; and to facilitate the exchange of research, intervention strategies, and information relating to fragile X syndrome (FXS), fragile X-associated tremor/ataxia syndrome (FXTAS), fragile X-associated primary ovarian insufficiency (FXPOI), and related forms of X-linked mental retardation.

This is a call for conference abstracts. Submitted abstracts will be accepted online beginning November 16, 2007, through February 29, 2008.

**ALL ABSTRACTS MUST BE
SUBMITTED ELECTRONICALLY.**

Please visit our website at www.FragileX.org, where you will find a link to the **Abstract Submission Form** under "Conferences & Webcasts" or by clicking on the conference logo. The foundation will respond directly to the first presenter listed on your abstract, to advise of acceptance or refusal. The presentation format options are PowerPoint, oral and/or poster. PowerPoint presentations can occur both within individual sessions and/or during the poster reception. All poster presentations may be displayed throughout the

conference and must be displayed during the poster reception on Friday evening, July 25, from 5:00 to 7:00 p.m. Presenters must be onsite during the poster reception. The foundation will provide mounting surfaces. Please note: **If you are presenting an abstract in any form, you must register for the conference.** (Student rates are available.)

Specific workshops will cover a broad range of disciplines and will be directed toward an audience of families, physicians, scientists, clinicians, and educators. Presentation topics can include a focus on pediatrics, molecular biology, genetics, cytogenetics, genetic counseling, educational issues and strategies, speech and language therapy, occupational therapy, legal and ethical implications, psychiatry, psychology, neurology, nursing, social work, pharmacology, or issues of specific concern for families, including national and international networking. Conference programs will be developed by the foundation in cooperation with its board of directors, conference planning committee, scientific and clinical advisory committee, and the membership. Conference format will include symposia, lectures, workshops, panel discussions, and poster presentations. All events are scheduled at the Hyatt Regency St. Louis at Union Station. Conference programming will run from 8:00 a.m. to 5:00 p.m. daily except opening and closing days. Social and leisure activities will be available in the evenings. ☎

11TH INTERNATIONAL CONFERENCE WORKSHOP TOPICS

<i>Academic Interventions</i>	<i>Financial/Insurance/Planning</i>	<i>Neuropsychological Studies</i>
<i>Adults and Aging</i>	<i>Gene Therapy</i>	<i>Occupational Therapy/S.I.</i>
<i>Animal Model Studies</i>	<i>Genetic Counseling Studies/Intervention</i>	<i>Premutation Carriers Studies/Issues</i>
<i>Augmentative/Alternative Communication</i>	<i>Growth and Endocrine Issues</i>	<i>Psychopharmacology</i>
<i>Autism and FX Studies/Issues</i>	<i>Inclusion/Mainstreaming</i>	<i>Reproductive Options</i>
<i>Behavior and Aggression</i>	<i>Medical and Neurological Issues</i>	<i>Screening & Prevalence</i>
<i>Studies/Intervention</i>	<i>Molecular and Cellular Studies in FX</i>	<i>Sexuality</i>
<i>Early Intervention/Preschool Programs</i>	<i>Molecular Pharmacology/Pharmacogenetics</i>	<i>Speech and Language Studies/Interventions</i>
<i>Electrophysiological Studies</i>	<i>Mothers/Fathers/Siblings/Grandparents</i>	<i>Spiritual/Faith-Based/Cultural Issues</i>
<i>Emotional & Behavioral Studies /Intervention</i>	<i>Neuroimaging</i>	<i>Synaptic Structure and Function</i>
<i>FMRP and Molecular Clinical Correlations</i>	<i>Multidisciplinary Studies/Interventions</i>	<i>X-Linked Mental Retardation</i>
	<i>Neurological Problems in Pre/Full Mutation</i>	



A Season of Overflowing Generosity!

Southern Wisconsin Golf Classic

Not to outdone by its previous successes, the 3rd Annual Southern Wisconsin Fragile X Golf Classic on August 13 was better than ever! Over 130 golfers joined PGA pro Steve Stricker for an outstanding day of golf at the beautiful Westmoor Country Club.



Jackson Pipkorn and his friend, sports news anchor Tom Pipines.

Many members of the Southern Wisconsin FX Resource Group teamed up to make this event a hole-in-one success. Key organizer Jeff Pipkorn expressed the group's gratitude to Steve for the gracious donation of his time, name and energy.

Golfers lined up for the opportunity to tee off against him, while kids enjoyed some one-on-one instruction.

Local sports news anchor Tom Pipines from FOX 6 Sports in Milwaukee welcomed guests to the dinner and a lively auction of some great donated items from local businesses and families.

Big thanks go to the event's generous corporate sponsors—Marriott Construction, Marking Services, Famous Footwear and Wahlgren-Schwein. A special thanks to the dedicated volunteers from the Southern Wisconsin FX Resource Group who contributed precious time and effort to make the event a huge success!



Tami Mueller and Julie Deming.



A group from corporate sponsor Marking Services is all smiles at the Southern Wisconsin Golf Classic.



Darienne and Laureen Majeski join Nina Liberati at the Michigan Golf Outing.

Michigan Golf Outing

It was a beautiful day for the 8th annual Michigan Golf Outing. There was no sign of rain—a very rare occurrence in Michigan come September! Despite the unusual conditions, 140 golfers and diners showed up for the fun. The day on the links included golfing, of course, betting (of course), and plenty of hotdogs, chips, pop and beer. Golfers entering the four closest-to-the-pin contests helped raise additional money. A delicious BBQ steak dinner was served, accompanied by a raffle and door prizes. Event organizers Nina and Frank

Liberati want to thank all the wonderful people who come out every year to support this event. And we would like to thank Nina and Frank and all of the Michigan families who work so hard to make it happen!

Going Green With Phones

The Fragile X Resource Center of Western Mass has collected more than 250 phones over the past two years and turned them into “green” cash to donate to the NFXF.

Group leader Denise Devine explains, “We send the old cell phones and batteries to a company that recycles the parts. Typically we get \$3-\$5 per phone, depending on condition, and they pay for shipping. For our group, checking the collection boxes and shipping the phones out is the only work involved. Before I send any phones, I check each phone for the memory card and destroy it. All funds raised go directly to the NFXF. It’s an easy way to raise money and it helps the environment at the same time.”

If you have old cell phones ready for donation, please send them and the batteries (no accessories, please) to: Denise Devine, 106 West St., Hadley, MA 01035.

Splash for Cash

More than 20 families, including grandparents, teachers and friends, joined the splashy fun in San Diego this July. Live music was provided by the Big Blues Band, a group comprised of individuals with Fragile X. A pool, water slide, trampoline, raffle and delicious lunch provided “fun in the sun” for all. Our thanks to the Fragile X Center of San Diego for its generous donation of funds raised from this event!

First Carolina Gala

The Carolina Benefit for Fragile X premiered on Saturday, September 15 in Cary, North Carolina. Scheduled in conjunction with a meeting of the NFXF Board of Directors, the event was co-hosted by Don and Pam Bailey and Joanne and Barry Roberts. The new Umstead Hotel and Spa (www.theUmstead.com) located on the campus of SAS Institute, provided a lovely venue, starting with an outdoor reception on the terrace overlooking the lake on a gorgeous fall afternoon.

More than 220 people enjoyed the festivities, capped off by entertainment from local author, musician, and humorist Clyde Edgerton. A long-time friend of Don and Pam, Clyde has written eight novels and one nonfiction book about his



Nathaniel Bailey & mom Pam Bailey.

adventures as a pilot. Drawing on his experiences growing up in the south, he kept the audience laughing with readings from his books, along with great music.

The silent auction also had a southern theme, including beautiful pottery from local potter, Ben Owen. Barry Roberts led the live auction with great gusto! The highest selling live auction item went for nearly \$5,000, but there is a story to tell about it. Clyde offered to auction the opportunity to have a bidder’s name or the name of someone they designate appear in his next novel! Two couples kept the bidding going up and up, until finally Barry suggested that Clyde include both names, to which he immediately agreed. Then, an inspired Scott Canel, a member of the NFXF Board of Directors, stood up and stated that he would match both of those bids if Clyde would also include the names of Don and Pam, the co-hosts!

The event was planned in only three months, and Don and Pam say that among the many things they learned with this first effort, one of the most important is to START EARLIER! They also emphasized how such an event cannot be pulled off without the help of many individuals. Nonetheless, it was a great evening and a financial success for the foundation. More important, it helped raise awareness about fragile X syndrome in the community and laid the groundwork for even bigger things to come.

continued on page 30 →



Some of the more than 220 people who enjoyed the Carolina Gala.



Looking Back on a Volunteering Year

“If we all did the things we are capable of doing, we would literally astound ourselves.”

—Thomas Alva Edison

I have this quote over my desk, and I think of it often as I communicate with our many wonderful volunteers every day. As I look back over this past year, I am “literally astounded” by all that they have accomplished on behalf of the foundation and the entire Fragile X community. It is an honor and a privilege to work with so many talented and committed individuals; their accomplishments serve as an inspiration to us all. My heartfelt thanks to everyone who worked so hard to realize their dreams!

'FUN'DRAISING EVENTS

St Louis Mouse Races—Rich & Dawn Erker and the FX Center of Missouri, St. Louis, Missouri

Coin Collection Competition—Students of McLean Middle School, McLean, Virginia

Delta Book Project—Mark Earnest, Holly Roos and the Central Illinois FX Resource Group



The St. Louis Mouse Races Mouse-Auctioneer Rich Erker.



A percentage of proceeds from this book go to the NFXF!



John, Winston and Jennifer Keith at Fashion Funds for Fragile X.



Holly Roos walks with son Parker, Congressman Phil Hare, and daughter Allison.

Matt Rhodes Run—Birmingham, Alabama

Raising Green in the Desert—Joan & Jay Canel, Judy & Irv Gross, Scottsdale, Arizona

Half Marathons for Mitchell—David Frey, Virginia Beach, Virginia

4th Annual Walk for FX—Holly Roos and the Central Illinois FX Resource Group, Canton, Illinois

Fashion Funds for Fragile X—Jennifer Keith, Roanoke, Virginia

Poker Run and Walk for FX—Burgandi Dotter and the Utah FX Association, Vernal, Utah

Fragile, Not Broken: A Fun Run/Walk for Knowledge—Diane Southard, Fragile X Center of Missouri, St. Louis, Missouri

William & Enid Rosen Research Fund and Marlyn & Melvin Weingarden Family Fund Virtual “Event to Remember”—Arlene & Jeffrey Cohen and Cindy & Melvin Schwartz, West Bloomfield, Michigan



Cheryl Cooper, Lynda Canel, Rebecca Fishman, and Jim Schallman at the Chicago Gala.

7th Annual Chicago Gala—Lynda & Scott Canel, Cheryl & Carey Cooper, Rebecca & Ira Fishman, Mary Beth Bohm and the FX Association of Chicago

Oak Leaves Book Launch Party—Maureen Lang Schmidgall, Libertyville, Illinois

Rummage Sale—Lynda Rogers, Becky Cox and the TeXas FX Association, Irving, Texas

CJ's 1st Annual Golf Tournament—Kevin Lotspeich, Concord, California

Brody's Birthday Bash—Wendy & Bubba Griffin, Lewisburg, Tennessee

Fragile X'treme Rock Concert—Julie Ditkof, Hartland, Wisconsin

Walk for FXS—Jennifer Mathias, Bernville, Pennsylvania

South Florida Dinner & Auction—Randy & Les Green, Ft. Lauderdale, Florida

Silverton Botox Benefit—Dr. Kimball and Jennifer Levine Silverton, Grand Blanc, Michigan

3rd Annual Southern Wisconsin Golf Classic—Jeff Pipkorn and the So. Wisconsin FX Resource Group, Brookfield, Wisconsin

3rd Annual Maine Motorcycle Ride for FX—Julie Gosselin and the Maine FX Resource Group, Gray, Maine

Voices for FX—Michelle Rocker and the Virginia FX Association, Charlottesville, Virginia

1st Annual Mayor Domenick Stampone Charitable Foundation Golf Outing—Bill Close, Domenick Stampone and Paula Fasciano, Franklin, New Jersey

8th Annual Michigan FX Golf Outing—Nina & Frank Liberati and the FX Association of Michigan

Carolina Benefit—Don & Pam Bailey, Cary, North Carolina

Jodi Feinstein Run—Chicago, Illinois

Maryland Golf Classic—Michael Kelley and the Maryland FX Association

3rd Annual San Francisco Gala "Night at the Copacabana"—Jane Jones, Sydney Rainin-Smith and the Northern California FX Association, San Francisco, California

Lizzie's 3rd Annual Open House—Nancy Reilly, St. Charles, Illinois

EDUCATIONAL SEMINARS—CO-SPONSORED BY THE NFXF

Fragile X & A Sensible Approach to Behavior—The Central Illinois FX Resource Group & Holly Roos

14th Annual Fragile X Syndrome Symposium—The Fragile X Resource Center of Missouri & Margaret Israel

Success with Fragile X Syndrome—The Fragile X Association of Southern California & Naomi Star

X-pert Advice: Understanding and Guiding Behaviors in Fragile X Syndrome—The Fragile X Association of Michigan and Kim Young



Marcy and Jeff Pipkorn at the Southern Wisconsin Golf Classic.



Lizzie at her Annual Open House.

Akron Children Hospital FXS Conference—Kristi Atwood and Cindy King

FX Educational Seminar—The Fragile X Resource Group of Greater Cincinnati-Northern Kentucky & Joe Garera

Are you inspired? Please call or email me to discuss your event plans for 2008! ☎

Linda Sorensen is the development coordinator for the NFXF. Email: Linda@FragileX.org



Mitchell Christoff and David Frey

Half-Marathoning for Mitchell, Part II

As some of you may recall, David Frey started the year with an ambitious plan: run two half-marathons, six months apart, and to run the second faster than the first. Although David was a little disappointed by his September race results, the screams of support from Mitchell Christoff provided wonderful consolation. Here is David's first-person account:

"Sunday morning, 6:45 a.m., and approximately 20,000 participants were lined up at the starting line in Virginia Beach to tackle 13.1 miles. An amazing site! There were so many runners whizzing by that an overwhelmed Mitchell Christoff had to sit down and take a break from cheering.

"The elite runners finished in an hour and three minutes...I never saw them. Mitchell did scream "Run faster, Uncle Dave!" at mile marker 9, and again near the finish line, which was great inspiration. (His screams are quite recognizable!) I lumbered in at 1:58.57—not quite as fast as my March race, but given that it was about 45 degrees warmer this time, I consider the day a success!

"The envelopes I've opened over the past eight months have been an amazing inspiration for everyone in our family. Your thoughtfulness and generosity have been truly heartfelt, and the Frey and Christoff families truly thank you."

Voices for FX

Five a capella singing groups recently performed at the 2nd Virginia Fragile X Association "Voices for Fragile X Benefit Concert." The groups included Academical Village People, Hoos in Treble, Silhoo'ettes, and Virginia Belles, all from Univ. of Virginia, and Sirens, from Charlottesville High School.

We deeply appreciate the compassion and support these young adults demonstrated by donating their time and talent to help raise awareness and money for Fragile X. In addition to

the vocal performances, the evening featured a raffle of items donated by local businesses, friends and family, including a tin of cookies baked by a young man with fragile X syndrome.

Numerous people donated their time and energy to make the evening a success. Thanks go to the Bankovs, the Carvers, the Dolans, the Keiths, the Matthews and Wilks families, and to the volunteers from the local Sam's Club.

Among those who donated \$100 or more: Dominion, Spectacle Shop, Dips & Sips, Crutchfield, TS Eways, Judy B., Dr. David Reynolds, Dr. Peter Toman, Dr. Kathryn Cook, Kathy Moore, Penny and Arthur Paikin, Kristin and Rob Carver, and Jennifer and John Keith.

Michelle Rucker, president of the Virginia Fragile X Association and event organizer, was very pleased. "When the idea of an a capella benefit concert was first born, the intent was to have a fundraiser that our children with FXS could enjoy and participate in without it being too overwhelming. For my own child, it is amazing how he takes to music. But, of course, music can be very overstimulating if played too loud. Fortunately, a capella music seems to fit the necessary criteria: it is calming, not too loud, but also fun. I made sure to let the audience know that dancing in the aisles and jumping up and down were acceptable behaviors at this concert. Of course, our children with fragile X syndrome were leading the way."

Maryland Golf Classic

The 3rd biennial Maryland Fragile X Golf Classic was held at Argyle Country Club in Silver Spring, Maryland on October 10. Over 140 golfers and volunteers enjoyed a beautiful 70-degree day of great golf, live and silent actions and dinner. Launched in 2003, the Classic has netted over \$300,000 for Fragile X. The winning low gross (actual) score of 11-under par was by the PMG ("Silver Sponsor") team of CEO Rick Powell, Ben Hiatt, Willie Brennen and Pat Price. The "Signature" sponsor, Bart and Associates, fielded two four-somes and had a third place net winner. Principally driven by just six dedicated families, this event has proven that even a small group of dedicated parents can make a big difference. ☺

Lynda Canel and Lisa Kelley chair the Development Committee of the NFXF Board of Directors. To learn more about supporting the NFXF, please contact Linda Sorensen, development coordinator in the foundation office: Linda@FragileX.org

Happenings 2008

A listing of upcoming educational and fundraising events for Fragile X.

Jan 12 Screening of documentary, "Living with Fragile X,"
1205 Soquel Ave., Santa Cruz, CA. *Info: ThinkMedia*
(831) 8459-7338.

Feb 26-27 2008 Advocacy Day, Washington, DC.
Info: NFXF office (800) 688-8765

April 18 California Fragile X Awareness Day, Sacramento, CA.
Info: NFXF office (800) 688-8765

19 5th Annual Walk for Fragile X Syndrome, Canton, IL.
Info: Holly Roos (309) 647-4864 or
roosfam@sbcglobal.net

May 10 Eighth Annual Chicago Gala, Chicago, IL.
Info: Lynda Canel (847) 433-9093 or Incanel@aol.com

July 23-27 11th International Fragile X Conference,
St. Louis, MO. *Info: NFXF office (800) 688-8765*

For more information or updates, visit
www.FragileX.org/calendar.htm, or call the
NFXF at (800) 688-8765.

Payroll Deduction Options Benefit NFXF

Supporters of The National Fragile X Foundation have two different ways to direct funds to the organization as payroll deduction enrollment periods commence. Thank you very much for considering these options.

THE 2007 COMBINED FEDERAL CAMPAIGN

The Combined Federal Campaign (CFC) solicits over 4 million civilian and military employees each year to participate in a payroll deduction plan to support their charities of choice. The National Fragile X Foundation is an approved charity of the CFC, and we greatly appreciate the ongoing support we receive from this campaign.

Please encourage family and friends who are federal employees to choose The National Fragile X Foundation as their charity of choice during the fall enrollment period. It's an easy and convenient way to support our mission of helping families impacted by Fragile X, and to fund research for improved treatments and an eventual cure. Thank you!

Note: The National Fragile X Foundation is registered with the Children's Medical Charities Federation under "Children's Inherited Brain Disorders Foundation." CFC #11129.

2007 UNITED WAY PARTICIPANTS

The support we receive from regular donors through the United Way payroll deduction plan also helps support our mission. Please consider choosing The National Fragile X Foundation as your charity of choice during this fall's enrollment period. Just enter the name National Fragile X Foundation on the enrollment form. Thanks!



The National **Fragile X** Foundation

BOARD OF DIRECTORS

President

Steve McKee
Wisconsin

Vice-President

Carey Cooper
Illinois

Secretary

Arlene Cohen, ASID
Michigan

Treasurer

Marty Lang
Wisconsin

Past President

Jeffrey S. Cohen, Esq.
Michigan

Don Bailey, PhD
North Carolina

Lynda Canel
Illinois

Scott Canel
Illinois

Todd Grayson
Illinois

Mark Gruzin
Maryland

Randi J. Hagerman, MD
California

Gail Heyman
Georgia

Margaret Israel
Missouri

Lisa Kelley
Maryland

Serena Lowe
Washington, D.C.

William Parker
Pennsylvania

EMERITUS DIRECTORS

Harris Hollin

Gerald McCabe

BOARD of TRUSTEES

John Harrigan, Esq.
President

Jay & Joan Canel

Joseph A. Christoff &
Catherine Frey

Cheryl Kraff Cooper, MD

Phillip & Sandra Cooper

Ira & Rebecca Fishman

Greg Fox

Manus Kraff, MD

Marty & Terri Lang

Norman Rosen

Carol Swig

Gene Warren

EXECUTIVE DIRECTOR

Robert M. Miller

SCIENTIFIC & CLINICAL ADVISORY COMMITTEE

Len Abbeduto, PhD, Co-Chair
Waisman Center, University of Wisconsin
Madison, Wisconsin, USA

W. Ted Brown, MD, PhD, Co-Chair
Institute for Basic Research
Staten Island, New York, USA

Elizabeth Berry-Kravis, MD, PhD
RUSH University Medical Center
Chicago, Illinois, USA

Martine Borghgraef, PhD
Center for Human Genetics
Leuven, Belgium

Marcia Braden, PhD
Private Psychologist
Colorado Springs, Colorado, USA

Jonathan Cohen, MD
Centre for Developmental Disability
Melbourne, Australia

Kim Cornish, PhD
McGill University
Quebec, Canada

Brenda Finucane, MS
Elwyn Training and Research Institute
Elwyn, Pennsylvania, USA

Paul Hagerman, MD, PhD
University of California School of
Medicine, Davis, California, USA

Randi J. Hagerman, MD
The M.I.N.D. Institute
University of California at Davis, USA

Gail Harris-Schmidt, PhD
Saint Xavier University
Chicago, Illinois, USA

Andrew Hoogeveen, PhD
Erasmus Medical Center,
Rotterdam, Netherlands

Frank Kooy, PhD
University of Antwerp, Belgium

Andrew Levitas, MD
University of Medicine and Dentistry
Stratford, New Jersey, USA

Allyn McConkie-Rosell, PhD, CGC
Duke University Children's Hospital
Durham, North Carolina, USA

Thomas Musci, MD
San Francisco Perinatal Associates,
San Francisco, California, USA

David Nelson, PhD
Baylor College of Medicine
Houston, Texas, USA

Christopher Pearson, PhD
The Hospital for Sick Children,
Toronto, Canada

Feliciano Ramos, MD, PhD
University of Zaragoza
Zaragoza, Spain

Stephanie Sherman, PhD
Emory University School of Medicine
Atlanta, Georgia, USA

Gail Spiridigliozzi, PhD
Duke University Medical Center
Durham, North Carolina, USA

Vicki Sudhalter, PhD
Institute for Basic Research
Staten Island, New York, USA

Jeremy Turk, DCH MRC
St. George's Hospital Medical School
London, England

Karen Usdin, PhD
National Institutes of Health,
Bethesda, Maryland, USA

Stephen Warren, PhD
Emory University School of Medicine
Atlanta, Georgia, USA

The National Fragile X Foundation
P.O. Box 37
Walnut Creek, CA 94597

Nonprofit Org.
US POSTAGE
PAID
Jefferson City, MO
Permit No. 210