What Is Fragile X?

The term “Fragile X” represents a group of genetic disorders, referred to as “Fragile X-associated Disorders,” that have a variety of impacts on affected individuals and their families. The disorders 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.

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**SERVICES OFFERED:**

Telephone consultation and basic informational packet—free

Educational resources (books, videotapes, CD)—fee

Local, national, and international conference sponsorship

Referral to medical, genetic and support services—free

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

**Phone, Email and Postal Contacts:** Approximately 10,000 annually

**Unique Website Visitors:** Over 600,000 annually

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

**Federal Tax ID Number:** 84-0960471

**Funding:** Individual contributions, family and corporate foundations, government grants

**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.
Movement, as much as anything, defines us. To be human is to move. And our overwhelming, relentless desire from the time we emerge from the womb is to have that movement be forward—toward new vistas, new discoveries, new opportunities for growth and expansion and fuller, richer lives. Beholding our children with fragile X syndrome, we sometimes see this compulsion toward movement play out even more dramatically than we would like, but (once everyone’s safety is assured!) we can’t help but admire and applaud the sheer tenacity they exhibit in the face of often imposing obstacles. Forward indeed!

In this holiday season, with your National Fragile X Foundation unveiling a new logo, a new slogan, a new website and a host of other new items still to come, forward is the identifying characteristic we seek to imbue into all our endeavors as another year beckons. Holding the riches of our 27-year history close to ourselves, we hail the children among us and the children within us, too, as we forge ahead—eyes ever on the prize of a generous and committed community, ever on the move...

HAPPY HOLIDAYS. HAPPY 2012.
A Fond Farewell and Appreciation

For the last four years I have served as vice president and then president of the NFXF. It has been an honor and privilege to serve on a board so rich with dedicated and talented individuals. We have made tremendous strides in moving the mission forward. Under the guidance of then President Don Bailey we were awarded the grant for the Fragile X Clinical & Research Consortium (FXCRC), one of the most important projects in the history of the NFXF. The consortium continues to thrive, having just received a second award that will carry us forward for the next four years.

We have significantly strengthened and expanded the board’s committee structure, thus broadening the pool of individuals helping to develop the strategies driving our mission. The LINKS network has also been strengthened and is providing increased support for numerous FXCRC clinics. We have moved forward into the 21st century with NFXF Facebook, Twitter, FXTAS.org, and have recently redesigned www.fragilex.org.

In 2009 the board instituted new term limits, resulting in the largest rotation of board membership since our inception. While the process certainly caused many of us great anxiety in the first two years, our current board is comprised of an extraordinary mix of individuals bringing new energy, new ideas and proven results. It is an exciting time for the NFXF board.

This year marks the end of the six-year term for Board Member and Treasurer Marty Lang. In the long history of the NFXF it would be difficult to identify any board members who have contributed more of themselves. His tenure as treasurer has provided unparalleled improvements in our accounting systems, budgeting, and financial management strategies, and significantly improved the entire board’s understanding of our fiduciary duties.

As someone who thrives behind the scene and avoids drawing attention, Marty has quietly played a major role in guiding the foundation through significant changes. He chaired not one, but two new committees formed in 2010. First the NFXF Clinics Consortium Committee—formed to evaluate the foundation’s role in the consortium and provide counsel to our executive director on FXCRC strategies. And most recently the Finance Committee, Marty’s brainchild, formed to develop fiscal strategies and review revenue and expense performance against budget.

Marty’s work with foundation staff has had a profound impact on the development of our standard operating procedures. NFXF Associate Director Linda Sorensen shared the following:

“What’s top of mind for you today Linda?” is how Marty frequently starts our calls. It’s a great way to get focused on what we need to be talking about, and I have always appreciated the thought behind that question. Four years ago I would have laughed if someone had told me I would be spending significant time on the foundation’s finances. However, under Marty’s patient guidance, I am now comfortable with some basic accounting procedures, enjoy planning and managing the annual budget and preparing for our audits, and have even learned a few tricks in QuickBooks. He has never questioned my ability to do all this but just encouraged me to learn what I needed to get the job done. It has been a terrific learning opportunity to work with Marty these past years. I hope to honor his trust in my abilities by continuing to expand my knowledge and expertise in this area.

Finally, Marty has served as a trusted advisor and counselor to three NFXF presidents. His professional manner and calming demeanor have provided a steadying influence to the Executive Committee and full board meetings. He is an astute listener who speaks only when he has something of value (or humor) to contribute. He constantly reminded us of the joy he experiences in socializing with FX parents and researchers at board meetings and conferences. His Midwestern charm put everyone around him at ease. He is a gentleman in every sense of the word, and I am honored to call him my friend and proud to have served with him on this board. The Fragile X community owes Marty a great debt of gratitude. The board will miss him sorely, but the legacy of his work will live on.

As I prepare for my next chapter as the “former” board

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Forward (With Image and Substance)

Hopefully you noticed our new logo and tagline “Forward,” produced on our behalf at no cost from professionals whose business it is to think about “image.” Image, of course, is important. So is the substance that supports that image. The word “logo” comes from the Greek word “logos,” which has had many meanings including “an expectation” and “order and knowledge.” Which is why we are pleased to announce that, in addition to a new logo (image) a completely new website (substance) at www.fragilex.org will soon be debuting.

We know that most of you get a great deal of your information from the web. Don’t most of us?! When it comes to Fragile X, there is no doubt more information available than anyone could absorb in a lifetime. That’s one reason why all of the staff, the webmaster, a web designer, our editor and the parents on the board’s support and education committee spent a full year thinking about and working on the new site. The questions they pondered long and hard included: How easy is it to find information? Is it understandable to the average person? Is it the best and most current information? Does each particular topic cover the full life span? Those are just some of the questions they asked themselves and each other. (Be patient, as it may still take a few months to update all of the content on the old site and bring it on over.) We’re excited about the new site, especially since we think it will make your lives a bit easier, which is, after all, at the core of our mission.

So, our logo may have changed, as well as the look and feel of our website (though not our web address). But our mission-driven commitment to you stays the same.

A Backward Nod to Those Who Started the Forward Motion
As we look forward, it never hurts to glance back, which is exactly what I had the chance to do when I attended a November gala in Denver in honor of NFXF cofounder Dr. Randi Hagerman. There, I was joined by many parents and professionals who played key roles in starting up the NFXF in the early 1980s, when it was a Denver-based organization. I also had the pleasure of giving a hug to Gene Koelbel, who, along with her husband Walt, has given generously to the NFXF over many years (especially during the critical early years and in later periods of great financial need). Though not parents of children with fragile X syndrome, Gene and Walt have always appreciated the importance of providing resources for families who have children with special needs. Most fortunately for all of us in this Fragile X community, they have always seen the NFXF as one of those resources.

Coincidentally, the gala took place the day before the unveiling of our new NFXF logo—a perfect closing of the loop!

Clinical Trials
You may have noticed that the NFXF is helping Fragile X clinics recruit for trials of new medications. We are thrilled to be able to help various pharmaceutical companies with this effort. So, whether you are reading a recruitment flyer on our website or Facebook page, or receiving one in the mail or your email in-box, please pass it along to others you think might be interested. There may be a short-term problem recruiting enough participants for all of the trials by all of the companies, but, in the long-term, this is a great problem for the Fragile X community to have, because it means many companies are studying a number of drugs, and thus many possibilities open up for improvement in the lives of those with fragile X syndrome.

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In the face of Fragile X, the time has come for us to act, to define ourselves and to make things happen. We all strive for a better life for our family members impacted by a Fragile X-associated Disorder. Scientists and researchers have been hard at work since the FMR1 gene was first discovered in 1991. We are all aware that real progress is being made on the scientific front. Many of us know families who are already participating in clinical trials of new drugs, and preliminary reports suggest that we may really be onto something. The autism, Down syndrome and developmental disability community at large have all taken note of the dramatic progress made in unlocking the secrets of FX, and many believe that correcting the interference with normal communication between neurons may truly change lives.

But we also find ourselves at a very difficult time in the life of our country. A persistently poor economy and political polarization seem to have brought us to a standstill. Agreement and middle ground seem beyond our grasp, and as a result, the truly remarkable progress made both in science and in services for those living with a condition like Fragile X is at risk of stagnation or even decline, given the drastic budget cuts being discussed in Washington.

As a nation we’re struggling to get by with less and to set priorities for the dollars we have. Those in power are charged with setting those priorities, but we need to remember—and remind them—that they represent us. We must communicate to them what is important to us and help them to set those priorities. If we fail to make our voices heard we run the real risk that our priorities will end up on the proverbial cutting room floor.

And so it is time for us to act, to define who we are and to make things happen. Join us in Washington, DC on March 6-7, 2012 for the 9th annual NFXF Advocacy Day. We’ve come too far to turn back now. We may not be able to expect increases, but we must speak against draconian cuts or even worse, elimination of funding for FX research and public health efforts. Join us; you’ll be the better for it.

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More info: advocacy.fragilex.org
Boston Clinic Serves Northeastern Hub

Founded exactly in stride with the formation of the Fragile X Clinical & Research Consortium (FXCRC) in July, 2006, the Fragile X Program at Children’s Hospital Boston draws patients from all over the tightly woven northeast corridor of the U.S., which includes Connecticut, New Hampshire, Vermont, Maine, and portions of Rhode island and New York that may be closer to Boston than the Fragile X clinics in those states. True to the form of most consortium clinics, Boston is affiliated with both a university and hospital setting, in this case the highly regarded Children’s Hospital Boston and Harvard Medical School. Given its Children’s Hospital affiliation and location, the vast bulk of the Boston clinic’s activities focus on evaluation and treatment of fragile X syndrome in children. The program also offers initial evaluation and referrals as appropriate for adults who may carry the Fragile X gene and be subject to fragile X-associated tremor/ataxia syndrome (FXTAS) or fragile X-associated primary ovarian insufficiency (FXPOI). Close working relationships with multiple nearby providers for those conditions ensure that families affected by any of the Fragile X-associated Disorders can readily access needed care, close to their home.

Six core members form the staff of the Fragile X clinic, led by the program director and clinical geneticist Dr. Jonathan Picker, MD, PhD, and clinic coordinator Sharyn Lincoln, MA, CGC. Clinics are conducted twice monthly by appointment, with wait times ranging from mere days on occasion to several months when demand is high.

In common with other Fragile X clinics in the FXCRC, the program provides a comprehensive evaluation involving multiple providers. “Families are seen over a two-day period that generally totals about seven to nine hours of active assessment and evaluation,” says Lincoln. “The first day requires about six hours, then patients return the next day for one to three hours and are done by noon. Spreading the visit over two days reduces the stress on the children and makes for a happier experience for everyone. Families from outside the immediate area often use it to spend a little time in Boston.”

The core assessments are provided in four main areas by specialists in:

- Developmental pediatrics
- Genetic counseling
- Psychology
- Occupational therapy

Depending on the findings of these assessments, referrals may be made to a wide array of other specialists affiliated with the program, including speech and language, psychiatry, seizure disorders, orthopedics and other services. “Families and primary care doctors receive very detailed reports that include clinical notes, genetics notes, and summary recommendations for behavior and occupational therapy activities that they can take to their schools, their pediatricians, and others involved in a child’s care,” says Lincoln.

Given the intensive program, only four families are scheduled per month. Many families return annually for the simple reason that “things change,” Lincoln says—“especially when the report’s recommendations are followed and children are receiving services that see them make the progress they are capable of.” Patients may also be seen by one or more providers for specific treatments in between formal program visits.

The educational program at the heart of the clinic’s mission is reflected in team members having been deeply involved in developing the new FXCRC clinical guidelines. Picker chairs the consortium’s clinical committee, and a number of the guidelines were authored by the Boston team.

On the research front, the Boston Fragile X clinic is preparing to join the collaborative studies for the very promising targeted drug trials for Fragile X. The program has also been participating in a study on the function of nerves in the brain, comparing a group of people with fragile X syndrome to another control group without the condition. The study uses transcranial magnetic stimulation (TMS), a non-invasive tool that allows researchers to examine nerve activity and function in the brain. For more information on the Fragile X Program at Children’s Hospital Boston, call (617) 355-4697, or log on to: www.childrenshospital.org/clinicalsites/site2242/mainpageS2242P0.html.
The Fruits of a Community’s Long Labor

This column usually focuses on behavioral issues and then offers suggestions or remedies based on research and experience. This issue, however, will divert slightly from that format. Instead, we will celebrate the fruits of many people’s labors by highlighting successes from some very special people who have fragile X syndrome (FXS). As I reflect on these successes and the history of my involvement with these individuals and those who have supported them, I am filled with excitement and enthusiasm. I am in awe of the energy, dedication and devotion they have shown through the years.

When I think back on my own professional experiences, there was a time when we were working hard simply to identify those affected by FXS and in some way quantify what we were seeing in them as a group. We were occupied with defining phenotypes and developing intervention strategies that were effective. This was important work, and it established the foundation for further study of the condition and its clinical underpinnings. But it was not nearly as exciting as the present. The hard work done by many back then has resulted in a number of wonderful stories that I want to share in this column.

Two years ago a couple brought their 8-year-old son with FXS to me to evaluate. They had read about effective educational strategies in the literature and wanted to access those strategies for their son. They knew he was capable, but he had not been able to learn how to read. They sought out intervention strategies to increase his probability of success. The evaluation enabled them to advocate for certain supports and teaching methods in his school. As their efforts grew, the district found a gifted teacher for their son and afforded her telephone consultation with me to develop additional interventions as needed. The young man is now reading and spelling better than anyone had anticipated. He is included in a general education classroom and is learning to make friends—all because two parents and a teacher joined together to accomplish great things!

Across the country on the west coast, another family handed their young son with FXS drumsticks and a drum set. His father (a professional drummer) provided him with opportunities to drum with him and learn music. As this shy young man matured, he became more and more accomplished, eventually joining the marching band in his high school—capped by performing a solo concert. All of this was accomplished because his parents refused to believe he was incapable of achieving his goal to be a drummer and play in the band.

Another mother in the south contacted a district’s director of special education and requested that her son’s teaching staff receive training on best practices with those affected by FXS. She had faith that her son could learn more adaptive behavior even though he was struggling with significant self-injurious outbursts. She was convinced that with proper intervention, his behavior would improve and he could be included with typical peers. The director heeded this mother’s efforts and contracted for services to develop strategies and train the teaching staff. In addition, the staff was given the opportunity to conference with experts monthly by phone to continue developing interventions that worked to maintain the boy’s adaptive behaviors. Currently,
his success is continuing as he participates in activities with his typical peers and is thereby further motivated to reduce self-injury.

Four sisters in the midwest with FXS became motivated to make educational materials for others affected by the condition. They learned to cut and laminate, package and mail out the products. Their efforts have provided materials to hundreds with FXS. Their success was rooted in motivation to help others with their own condition, a foundation of support from their parents, and an overall sense of compassion.

A 40-year-old man in the west affected with FXS always wanted to learn to read. He was very aggressive and violent as a child and adolescent and was never emotionally stable enough to be educated with much success. His mother weathered a long storm of psychiatric placements and hospitalization but she never lost sight of his dream. As he grew older, she sought out private tutoring and for Mother’s Day last year received a DVD of her son reading to her. Imagine the impact this beautiful gift had on her—and her son. It was only possible because she refused to believe he was too old to learn and to accept that his potential faded with age.

A father from Germany attended the 2008 International FX Conference in St. Louis and heard about clinical and educational techniques that could change the outcome of those affected by FXS. He decided to take that information back to his country, where he worked for a year organizing an educational conference that reached an astounding 300+ teachers and parents in Germany. His dedication to his son prompted him to bring possibilities to others in his home country so that they too might experience success.

There are many more such stories of families and support workers who have been motivated by these very special people with FXS to push through brick walls and climb mountains supporting the development of their potential. I am humbled to have been among them and to call them colleagues and friends in this remarkable Fragile X community.

The author is a psychologist and special education consultant in Colorado Springs who specializes in Fragile X. She is the author of Fragile: Handle With Care, and creator of the Logo® Reading System. Contact: info@marciabraden.com

Write for the Fragile X Quarterly!

Finding yourself telling tales over your coffee cup about your kids and other family members living with Fragile X? Are you reflecting on a blog, private journal or social media site about your experiences as a parent of a child with FXS, or as a son or daughter of a parent with FXTAS or FXPOI? Please consider sharing those thoughts for the benefit of readers in the wider Fragile X community. Fragile X Quarterly editors always welcome and work with writers, from professionals to absolute beginners, who wish to explore the many dimensions of life in the Fragile X world. Writing opportunities include:

• Parents’ Forum
• In Their Own Words

No ideas or tales are too small or large. Send submissions or discuss your ideas with: editor@fragilex.org. Thank you!
realized when my son Ian was just 4 years old that he had difficulty with transitions. I had never thought about transitions being difficult. You just did them; you didn’t think about it.

Well, with Ian, I started thinking about them. I still do to this day. I quickly learned I had to allow plenty of time for everything, arrive early to events, set up schedules and routines, take time for transitioning back home—and that was just the beginning. Over time, the work required to make a transition successful for Ian became more complicated. And good or bad, thinking about transitions has become an automatic part of my life.

There are big transitions: moving to a new house or city, starting a new school. Then there are daily transitions: getting up in the morning, getting dressed, eating breakfast, getting off to school or work—basically every aspect of a typical day. Then there are special transitions: vacations, summer breaks—all scheduled change of the daily routine. And the unexpected transitions: fire drills, illnesses.

When Ian started kindergarten (a big transition) I chose the school I thought would be best for him, chose his teacher, educated the teacher, gave a presentation to the entire faculty, and talked to the students in his class and to parents I saw around the school.

For the daily transitions, I created a picture schedule of each day, and I helped develop strategies to transition between activities, such as prompting that an activity was about to end, mentioning what activity was going to happen next, using a transition object, and encouraging use of sensory activities.

For the special transitions of vacations and school breaks, I created a schedule with routine. I had to. These transitions were so difficult for Ian that I soon learned the first two weeks of summer and most of winter break would be filled with aggressive behavior, and many vacation trips had aspects of complete disaster—no matter what I did.

Unexpected transitions such as fire drills were successful because we did not let them become unexpected; we told Ian about them ahead of time. Even telling him sent his anxiety through the roof. When he wasn’t told about a fire drill, things went flying, and usually his glasses were the first thing to go.

So, what happened over time? I figured out better what Ian needed in order to make transitions successful. I still did/do a lot of the work, but it is from a base of experience, not figuring out on the fly what needs to be done.

The last big transition he went through? From school to out-of-school. I set it up so the last two years of school, his routine was: go to school, then reading class, then PE, go to work, then back to school to help out with the football team.

When he graduated, two parts of his day did not change: work and football. He got to sleep in late, go to work, come home for a snack and then go back to the high school to continue helping out with football team. He liked the sleeping-in-late part.

For his daily transitions:

- I figured out how much time he needed to make his different transitions throughout the day. For example, to get ready for work in the morning, he needs 75 minutes. Every now and then I try reducing it to an hour, but it doesn’t work—he is then 15 minutes late to his job. Basically, as long as his daily schedule has routine, predictability and plentiful time built in, he does fine.

Lucky for him (and me) he has a job with the same schedule each day and where he alternates between gross motor and fine motor activities. He works at our local grocery store, bringing in the carts and bagging groceries. It is a social job—he loves it and they love him. And every afternoon after work he has an activity through our parks and recreation department that is both physical and social in nature, such as basketball, softball and volleyball.
I tried different visual schedules, and over time Ian told me what he wanted: a calendar that hangs on the wall by our dining table that lists his daily activities. He wanted them hand-printed, and he did not want pictures, so I print the names of his activities. Since I know his reading is limited, I use short, simple words:

- Work 10-2
- Basketball 4-5

I have also learned how to talk to him about his day. When it looks like something is bothering him, I ask, “What are you thinking about?” rather than, “What is bothering you?” Often he brings up an activity he is anxious about. Then, using his calendar, we run through everything that is to happen during the time period that is bothering him.

I am more flexible. When Ian asks what we are having for dinner tomorrow night, I ask him what he wants to have. When he tells me, I tell him that is fine, but I also know he will probably change his mind, and I let him know that is O.K., too.

I praise him for a job well done, especially if I know the situation had him a little anxious and he was able to work through it.

SPECIAL TRANSITIONS

Because he is out of school now, we do not have the various breaks associated with the school year. So the “special” transitions revolve around vacations. In this phase of our lives, my husband and I spend most of our vacation time visiting our parents whose health is deteriorating. Ian likes to visit both sets of parents, so these transitions are much easier than they have ever been. That said, he does like to know what we are going to eat while we are there. Getting that resolved before we leave seems to “ground” him. (And he likes food.)

The other thing I can do now that I couldn’t when he was younger is leave him by himself for 1-2 hours. If the rest of the family wants to do something that he doesn’t interest him, I set him up with a snack and the Food Network on television. And he always has his cell phone handy.

The unexpected transitions are still difficult for Ian. But I have discovered that if something comes up and I allow him time to process and think about it, he often comes around. “Ian, we just got tickets to a CU football game. Do you want to go?” Ten minutes of thinking and he says, “Sure.”

I sometimes think about how Ian would respond if we had a fire, and the only approach I can come up with is, “Ian, there are some firefighters outside who would like to talk to you right now.” Hopefully this will never happen. But something for you to think about: I have gone to our local fire station and talked to them about the unknown of Ian’s response in an emergency. When our address comes up on their system, an icon tells them there is a young man with a disability living in the house whose behavior in an emergency is unknown. It also has the location of his bedroom.

The difference now for Ian? He understands more about what to expect when he goes places, especially ones he has been to before. If he has not been to a place, I compare it to something he knows. He also has a better concept of time (thanks to 30-minute Food Network shows). He is also able to tell me when he does not want to do something, and I respect that as often as I can.

A final thought: For the most part, “hurry” is not in Ian’s vocabulary. That is a term we should all strive to use much less.

The author is the support services coordinator for the NFXF. Send your questions to her at: treatment@fragilex.org
Helping Manage FXS in the Classroom

Students with fragile X syndrome often seem to be described like the mythical girl with the curl in the middle of her forehead: “When they are good, they are very, very good, and when they are bad they are...well, let’s just say, ‘quite disruptive.’” What accounts for our lovable, good-natured, eager-to-please angels morphing into Tasmanian devils quicker than you can say, “Fragile what?”

Teachers, many of whom are well-equipped to work with kids prone to demanding behaviors, find the unpredictability with which our charming cherubs create havoc especially perplexing. The most accomplished masterminds of consistent environments are foiled by students for whom antecedents are undetectable and consequences appear to be inconsequential.

It’s not that these children are impervious to tried-and-true behavior management techniques. At times, however, the neurological wiring characteristic of FXS results in an internal condition that supersedes environmental influences. The difficulties our children experience with self-regulation can overwhelm their ability to respond to their surroundings with intention or control.

We know from numerous research studies that when individuals with FXS are in a highly aroused state, it takes them longer to return to relative calm than it does unaffected peers. Acting-out behavior exhibited during such a state is no more a function of attention-seeking than is a panic attack. The strategies typically employed for treating inappropriate behavior—such as ignoring or imposing a conditional consequence—will therefore not be successful.

While we do not at this time have an anti-arousal power tool in our tool chest, we do have a number of potentially effective tactics we can employ. Calming techniques based on principles of sensory integration can assist agitated children in reorganizing their processing capabilities. Providing a safe and undemanding space, free of stimulation, can be helpful. Introducing a “sensory diet” throughout the day may prevent arousal levels from escalating. Medication can also help.

Teachers must contend with additional complications when addressing the range of challenging behaviors in kids with FXS. A child who is encouraged to go to a quiet space when showing signs of aggression can appear to be given an unfair pass when other children face consequences for what looks like identical behavior. School personnel may be unable to accept the application of a deep pressure massage to a child who is hitting and kicking as anything but unacceptable reinforcement. What this suggests is that educating our FXers requires educating others as well. There are times when members of this unique population call attention to their difficulties in no uncertain terms, which in turn calls upon their special education community to expand their efforts to understand them.

I am not suggesting that the answer to behavior problems in students with FXS is to routinely allow them to escape from demanding situations. Accommodating their needs for calming when upset must be tempered with strategies to help them learn to adapt to taxing conditions. The choice of strategy needs to be based on understanding what is motivating the behavior at any given time. Top-notch detective work is required from teachers of children with FXS. Is throwing a shoe an inappropriate request for attention, a sign of physiological discomfort, or a deliberate action employed to achieve a time-out? If it’s the latter, should time-out be allowed in order to address a need for a break from stimulation? Or should the student be guided to adapt to classroom expectations? Skilled observation, functional behavior analysis, and careful evaluation of strategy effectiveness can lead to a judicious course of action.

Recognizing to what degree an undesirable behavior is within a student’s ability to control is key. We know an FXS diagnosis means an altered physiology. This manifests itself in ways unique to each affected individual. Understanding how this influences a particular student provides teachers with valuable information that can be translated into an individualized program encompassing an educationally attuned, two-pronged compromise:

1. Accommodation on the part of the community within which the student is a member.

2. Adaptation of the student to the expectations of the community.

Knowledge of how fragile X syndrome affects a particular student will help the IEP team discern the parameters of this delicate balance.

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This past summer, I conducted research on fragile X syndrome (FXS) in the Center for Interdisciplinary Brain Sciences Research (CIBSR) at Stanford University. My project focused on improving our understanding of the neurodevelopment of young children with FXS. Since FXS is first seen in early childhood—a time of significant growth and development in the brain—understanding neuroanatomy at this age improves our knowledge of both abnormal and normal growth processes.

Before my arrival at CIBSR, the neuroscientists had used MRI technology to scan the brains of young boys in three groups: one with FXS, another with typical development (TD), and a third with developmental delay (DD). The boys were scanned twice in a two-year interval, first when they were between one and three years old and again when they were between three and five.

To analyze these MR images, I used an image analysis suite called FreeSurfer, a largely automated program that provides full brain information, including the volume, surface area, thickness, and mean curvature for each cortical region. Ultimately, I hoped to determine whether FreeSurfer is an effective tool for studying the neuroanatomy of young boys with FXS. If we establish its utility and validity in FXS, we could open the door to future research that will provide critical information for neurodevelopmental studies and clinical trials.

Since I had begun learning FreeSurfer during the school year, I quickly became proficient in the editing techniques necessary to guide its analysis of the MR images to completion. My task was to compare the neuroanatomy of the boys with FXS to those with TD/DD, then analyze how they changed over time.

My results were largely consistent with those of previous studies, and I concluded that FreeSurfer is an effective tool for the purposes cited. One of my main findings was that the boys with FXS had a significantly larger caudate nucleus than the boys with TD/DD. This result was important because the caudate nucleus serves in the brain’s learning, motor, and cognitive control system, and previous studies have shown that an enlarged caudate nucleus is a defining characteristic of FXS.

I also found that brain volume and thickness differences between the boys with FXS and the boys with TD/DD became more pronounced over time. The increase in brain thickness differences was especially interesting because neuroscientists have not examined thickness as much as gray or white matter volumes. Abnormal thickness may in fact play a role in the cognitive and behavioral impairments of individuals with FXS. More research is needed to further investigate this hypothesis.

During my time at CIBSR, I had the tremendous opportunity not only to collaborate with deeply committed scientists to increase our knowledge of FXS, but also to meet a young man with FXS and his father. This was the most rewarding part of my fellowship experience because I got to personally know the people whom my research could help. While learning about their lifestyles, accomplishments, and difficulties, I was inspired by their resilience and courage. Not once did they feel sorry for themselves; instead, they exuded an optimism and closeness that allowed them to overcome challenges most families hope not to face.

I will carry the scientific and personal lessons I learned at CIBSR to my future research and aspirations to become a medical doctor. I am extremely grateful to my mentors at CIBSR for teaching me about neuroanatomy and meaningful scientific inquiry, to the William and Enid Rosen Research Fund for granting me the fellowship support, and to the families affected by fragile X syndrome for helping us better understand the disorder.
recently finished my graduate thesis about fragile X syndrome, and it’s turned out to be more of a memoir than I had intended. But I shouldn’t be surprised. I decided to write on FXS because my brother has it, and it was an attempt to understand not just him better—at those two mysterious levels of molecule and mind—but also my family and, let’s face it, myself, forged as we all are by the strange pressures of that wayward gene.

My research revealed some fascinating things, not just about the way FXS affects brain development and passes down through the generations, but also about how important the FMR1 gene is to shaping consciousness in general—the self, if you want to get spooky about it. In studying this research, though, there’s one issue that seems to escape inclusion in descriptions of the phenotype, a quality that seems fundamental to me, and maybe to you as well: the Fragile X sense of humor.

My brother makes people laugh. He’s on the moderate-to-severe end of the impairment spectrum, shows all those quirky signs of autism, and still has trouble with toilets. But, man, can he sail a one-liner across the bows. He’s got a way with humor that is singular, surprising, and revelatory.

Now, it’s easy to suggest that maybe we’re all just laughing at what is only funny coming from a “poor retarded kid.” That we’re grading on a curve, so to speak. “Kids say the darndest things,” and all that. But I contend, and I suspect you’ll agree, that the humor that issues forth from people with FXS is brighter and sharper than so much of what we encounter in this often-dull world. It is absurd because its circumstances are absurd. The priest and the zebra forever walking into that bar perhaps also think, in their quiet moments, that their lives didn’t turn out quite how they planned.

Humor is, of course, a tricky thing to qualify or quantify; try to explain it and it stops being funny. It’s the birthday cake that slips away when it’s held too tight, the bicycle you crash when you start thinking about the mechanics of riding. It’s that pure thing that, like love, is only true when it’s felt rather than figured. This may be why research has such a hard time with it.

As it happens, humor has been studied for thousands of years by such notables as Plato, Immanuel Kant, Thomas Hobbes and Sigmund Freud. Most definitions of humor fall into one of three theories: Incongruity (in which an outcome is so unexpected as to shock us into laughter); Superiority (in which we laugh at the realization of our greatness over a person or thing); and Relief (in which humor is a way of releasing the repressed tension of a situation). No theory has offered a complete explanation of humor, though, so science has taken up the task. One researcher, Peter McGraw of the University of Colorado, Boulder, reveals in a May 2011 Wired magazine article his “benign violation theory”: Something is funny when it violates our perceptions of the world, but in ways which don’t fundamentally threaten that world.

Your correspondent feels that this theory rather sums up the daily experience of having FXS in a family: Every day, something new to free us from our desperately held perceptions. This will not be an opinion held by all those touched by FXS, who may see the violation without the benignity. It is your correspondent’s opinion that this is an attitude that may—in fact, must—change. Learning to appreciate the FX sense of humor is a start.

Having long observed my brother, I’ve identified many types of FX humor, of which I present three examples here. (I encourage you to take your own assessments, there at the dinner table or wherever your subject feels most inspired; there’s always additional research to be done.)

**Humor Type:** Wordplay; reworking familiar concepts in new ways

**Case Study:** The dog peed in his bedroom.

Mother: “Why did she pee?”

Brother: “Maybe her penis hurts her.”

Mother: “She doesn’t have a penis. Girls don’t have a penis. Girls have a vagina.”
Brother (thoughtful, gently surprised): “Oh. Oh.”
Mother: “So, not her penis, but her...vagina.”
Brother (having it both ways): “Vagin-is.”

**Humor Type:** The short con; a grift in which he tries to get what he wants by inventing facts/figures (and proves he’s smarter than he lets on)

**Case Study:** Going to a movie, something loud that will no doubt inspire him to rock until the whole row in the theater shakes.
Brother: “Can I get popcorn?”
Me (joshing): “I don’t know. You think that’s a good idea?”
Brother: “It is. It’s good for me. It’s good for my skin.”

**Humor Type:** Existential; alters perspective

**Case Study:** Our grandparents are deceased; this dialogue is part of the continuing attempt on my part to understand what this means to him.
Me: “What are Grandma and Grandpa doing?”
Brother (after a pause): “They’re just chillin’.”

There are other examples, tons of them. Many of them favoring slapstick, props and (I’m sure you won’t be surprised) bathroom humor. And maybe, yeah, maybe some of the humor is situational, maybe it’s the product of having a brother—or a son, a daughter, a sister, a crazy uncle—who isn’t ever going to be “normal,” and then you start to think about what “normal” really means anyway, and just the thought that everybody’s maybe trying for something we all call “normal” is itself absurd, and worth laughing at, and every day there’s a reminder of that absurdity in little comments, little gestures, and it’s reassuring to know that that’s all right.

And it had better be, since we in the FX community are formed in so many ways from those absurd experiences, whether it’s a shoe thrown across a restaurant, food grabbed out of a salad bar and tossed back half-eaten, or, say, taking one’s brother on his first airplane flight, in which he sees clouds out the window of the emergency exit row (which at the time one had to insist on if one cared to have any legroom, even though one has to assure the flight attendant that, of course, we are both sound and capable in case of emergency, even as the brother rocks and chews his knuckle and emits a high-pitched whine, and at which point one has to admit to oneself that, yes, the lives of these 250+ people are more than worth putting at risk so that one won’t have to be folded up like a Chinese acrobat for the 2+ hours from Atlanta to San Antonio). And then the brother declaring, at the top of his voice and right in the midst of the turbulent ascent: “Smoke!”

Anyway, this FX sense of humor is not just one man’s observation. Dr. Randi Hagerman of the University of California Davis MIND Institute, a resource and subject of my thesis, recognizes the phenomenon as well. “Kids with fragile X syndrome can have a great sense of humor,” she says, “and they are almost always happy. That’s a very good aspect of the phenotype.”

She also notices something else, other advantages of the phenotype: “They can be sensitive to interpersonal issues. And so in many ways they can be wonderful children for these families. These are unique kids.”

Still, in my research I found so many accounts, on blogs and in news stories and in conversation, of the hardship faced by families living with FXS. Tragedy, they sometimes call it (comedy’s evil twin). Depression and anxiety and guilt—all in there, too. The frustrations of everyday problems, like cleaning a child who should long ago have figured out how to use the bathroom. A lot of these stories are from parents with young kids, children really. My brother is 30 now, and while there are still times when the bathroom is an issue, it is easier and better and, with three decades of perspective, clearer to me that that other life, that imaginary “normal” life, isn’t the sort of thing I’d ever want. How many days are we willing to lose in desiring that which doesn’t even exist?

For parents and siblings who don’t see the humor in these trying days, consider this: What’s funny, what’s really funny, is to clean up after your adult brother and recognize that another pair of drawers gone bad is just another benign violation.

For the pursuit of a normal life and reveals a whole new kind of love, and duty, after the punchline.

The author grew up in San Antonio, TX, worked as a journalist in Austin and then in Richmond, VA, and recently earned his master’s degree in specialized journalism as an Annenberg Fellow at the University of Southern California in Los Angeles. He currently lives in Kansas City, where he says he’s considering how to turn his thesis “into something someone might actually be able to read.” Email: brandonreynolds@gmail.com
The Books and Bookends of Fragile X Life

Constructing a life around Fragile X is like collecting books. You keep the best, most important books securely within a good pair of bookends, and you toss the rest. In your home library, you may have 10 self-help books, some nonfiction, literature, and some fantasy and art books. Only some of the “books” that help you construct a positive life in the face of Fragile X might be actual books, while many others would be in the form of strategies on development and behavior, education, social skills, and the mechanisms of Fragile X itself. While the first few of these “books” may be dedicated to early development and skill acquisition, the titles become more specific and defined as the collection grows.

And let’s also not forget the bookends. The bookends of our “collection” are what hold the volumes of our knowledge firm and steady while allowing for expansion.

In a real bookshelf, the bookends are the structure, which might be whimsical and decorative. But in our Fragile X lives, the bookends signify structure in a more pivotal manner. The bookend on the left signifies everything we know about Fragile X and where we are now, and they inform us in selecting the books we might purchase. The right side bookend signifies the future, our end goals, which are dynamic, and which form in reference to the left bookend.

Why the bookend metaphor? Well, the privilege of accumulating over 20 years of experience working with families with fragile X syndrome has taught us many, many things. One of those is that keeping your eye on the future helps you establish and prioritize goals...now. In this way, the future informs the present, and vice versa. And it’s both the present and the future that allow you to build a meaningful collection of books, which is itself a reflection of a meaningful life.

But let’s get back to those bookends. The left bookend we conceptualize as the underlying characteristics that are common to Fragile X. We have learned that when you have this framework of understanding, your strategies, or your books on the shelf, become relevant and tailored to those specific characteristics. If you didn’t have this framework, your books would tend to be broad, general, off-base—not really useful. An example for our purposes: Knowing that your child is a visual learner who doesn’t understand sequences helps you to determine what reading curriculum the school might utilize for teaching this essential skill.

The right side bookend is equally important in constructing the framework. Visualizing the future opens possibilities while grounding you in the reality of what it will take to get there. This is where the future informs the present. For instance, if you envision your son with FXS working in the family business, what will it take to get him there? By reflecting on the left bookend, where you are now, you can begin to prioritize what your son will have to absorb in school and therapies and home routines (the right side bookend) in order to harness strengths and support areas that will help meet the goal of someday working in the family business.

Both bookends allow you to better utilize therapies, school programs, and community resources, in addition to knowing what additional books to buy that will fortify your collection. The books on the shelf are just as important as the bookends. In our metaphor, we think of the books as the strategies or means for achieving future goals. Our experience suggests that the collection should include several categories that together form the lifelong framework from which you construct an appropriate intervention plan.
This will allow you to create the supports needed to live the Fragile X way. The categories we suggest include:

1. Sensory issues and hyperarousal—This should extend to anything about the basic biology of Fragile X.
2. Basic behavior management—The take-home message here is you need to determine how to increase the likelihood that what you want to occur again will, and conversely, what you don’t want to happen again, won’t.
3. Routines and visual supports—These allow individuals with FXS to function at their optimum, and are essential tools for any intervention plan.
4. Task analysis—This allows for modification of the task structure, demand or environment so the individual can be successful.
5. Skill acquisition—What are the biggest challenges faced by the individuals and how can they be supported to acquire the highest skill within their potential?

Now, to extend our metaphor into more practical applications:

The left bookend is comprised of solid evaluations by professionals who know about Fragile X. The Fragile X Clinical & Research Consortium is a great resource for this (www.fragilex.org/html/links_groups.htm). The actual book by Dr. Randi Hagerman, _Fragile X Syndrome_, is an important reference.

The right bookend is established through some sort of “futures planning” process. There are formal methods that can assist in this effort, starting from a very well done IFSP or IEP to any one of the suggested resources below (from www.allenshea.com/resource.html):

- **PATH: A Workbook for Planning Positive Possible Futures.**
  Uses an eight-step process to help people figure out life goals, build their strengths, include others in a personal support network, and, develop a commitment to action. This booklet was written by Marsha Forest, John O’Brien, and Jack Pearpoint and is printed by Inclusion Press. Ordering information: Marsha Forest Centre, www.marshaforest.com, 47 Indian Trail, Toronto, ON M6R 1Z8 Canada, (416) 658-5363

- **It’s Never Too Early, It’s Never Too Late!** The goals of Personal Futures Planning are to help someone develop a picture of what the future will look like, to build a circle of people who will help support that picture or plan, and to take some first steps. For a copy of this booklet by Beth Mount and Kay Zwernik (1988), contact the Minnesota Governor’s Council on Developmental Disabilities, (877) 348-0505 toll-free, (651) 296-9962 TDD, admin.dd@state.mn.us, or www.mnddc.org.

- **MAPS (Making Action Plans).** MAPS helps bring together the key people in someone’s life to develop a support plan. A MAPS get-together is usually hosted by two people, one who helps guide the meeting and one who records what happens on wall charts or an easel. For more information, contact the Centre for Integrated Education and Community as noted two paragraphs above.

As for the “book collection,” here are some favorite categories that will hold you and your growing collection in good stead:

1. Sensory/Hyperarousal
2. Basic Behavior
3. Routines/Visuals
4. Task Analysis
5. Skill Acquisition

Accumulate solid, well-considered resources in those categories and you will be on the road to filling the space between your bookends in a way that will prepare your child in the best possible way for the future you are busily building today. Happy book (and bookend) hunting!

*Tracy Stackhouse, MA, OTR and Sarah “Mouse” Scharfenaker, MA, CCC-SLP, are co-founders and directors of the Developmental & Fragile X Resource Centre in Denver, CO. Email: tracy@developmentalfx.org, and mouse@developmentalfx.org*
Forward With Fragile X Researchers

Editor’s note: The Fragile X scientific community has been exuding an almost palpable sense of excitement in recent years as researchers and clinicians continue to build upon one another’s work to make steady advances in our knowledge of all three Fragile X-associated Disorders. This knowledge has resulted in improved interventions on virtually every front: medical care, education, physical and occupational therapy, behavior management, pharmacology, genetics, and more. The National Fragile X Foundation is privileged to count among its Scientific and Clinical Advisory Committee members many of the premier scientists and clinicians who are driving much of the progress in the field. We are pleased to hear from a quartet of them below about their plans for continuing to move Forward With Fragile X.

Frank Kooy, PhD
Professor and Chair
Cognitive Genetics Group
University of Antwerp
Antwerp, Belgium

Corrine Welt, MD
Associate Professor
Harvard Medical School
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It looks like 2012 will be the most exciting year yet for our seven-person group researching hereditary intellectual disabilities including fragile X syndrome. Over the past seven years, we have been working hard to identify processes that are disturbed in the cells of patients with FXS. Since the causative gene is involved in many processes in the cell, this has not been an easy task. In the brain, two pathways work together to make us who we are: the excitatory pathway (the accelerator) and the inhibitory pathway (the brake). From what we know now, FXS results from a disturbance of the delicate balance between both pathways.

While many research groups have worked to intervene with the excitatory system, we hypothesized that the inhibitory system is also disturbed in patients with FXS. Our research has clearly demonstrated that drugs that improve the function of the inhibitory system can prevent epileptic attacks in mice. We also demonstrated that the same abnormalities that were discovered in animal models are present in humans. We are thrilled that the same drug that we used successfully in mice will now be used to treat patients in upcoming clinical trials.

Women who carry the Fragile X premutation helped make the discovery that some seem to experience an earlier menopause than other women. We know that some women who experience infertility may also be predisposed to early aging of the ovaries, which leads to earlier menopause. Since 2008, our group has been working to determine if there is an increase in the number of women who carry premutations or gray zone/intermediate repeat lengths when infertility is related to early aging of the ovaries, compared to when infertility is related to other problems.

In early 2011, we found there were more women undergoing infertility treatment due to early aging of the ovaries and who carried the premutation or intermediate/gray zone repeat than women who had other causes of infertility.

In 2012, we want to start to figure out the smallest CGG repeat length that might affect aging in the ovaries. We also want to use a virus model in ovarian cells to see if we can determine why the repeat affects the ovary. Finally, we want to see how environmental exposures like passive cigarette smoke might affect the ovaries of women who carry the premutation. We hope to improve understanding of this understudied ovarian problem.
Research by the Willemsen laboratory will continue in both fragile X syndrome and fragile X-associated tremor/ataxia syndrome throughout 2012. We are pleased at our long historical association with Fragile X. In 1991, we were involved in the identification of the FMR1 gene as the causative gene in FXS. Three years later, the generation of the Fmr1 KO mouse now used as an animal model for FXS worldwide was established in our lab. Looking forward, the growing understanding of the molecular mechanisms of FXS has led to therapeutic strategies designed to reverse the intellectual and behavioral problems associated with the condition. In the forthcoming year, we will focus on translational research to test the efficacy of various therapeutic compounds.

On the FXTAS front, we will focus on understanding the molecular mechanisms underlying the condition. As a first step to develop molecular interventions and produce pharmacological agents to treat patients with FXTAS, we need to better understand the expectations for reversibility of the disease. Our lab will work in conjunction with the NeuroTherapeutics Research Institute (NTRI) at UC Davis in developing a new mouse model to establish the degree and timing of reversibility of neuropathology and behavioral deficits. State-of-the-art methods will be used to characterize these genetically modified mice at different ages in their development, from birth through their reproductive life.

Next year will be a very busy one for me, as I was recently elected the 2012 president of the National Society of Genetic Counselors. For decades, genetic counselors have been on the front lines with families as they sort through the complicated genetics of Fragile X while adapting to the emotional and familial impact of its associated disorders. Many new developments are taking place in the broader genetics world, such as the emergence of technologies that will greatly increase the number of people diagnosed with genetic conditions—including Fragile X-associated Disorders. These developments have important implications for the NFXF, for genetic counseling, the disability community, and the population at large.

On a parallel front, Elwyn and other centers will continue to be actively involved in clinical trials of targeted pharmaceuticals, as well as promoting special education and behavioral practices that enhance the quality of life for children and adults with fragile X syndrome. Over the next few years, the convergence of these two developments—a significant increase in diagnoses plus advances in treatment—will have a profound effect on our Fragile X community. I look forward to continuing my ongoing work with Fragile X during this exciting time.

A Fond Farewell... continued from page 2

president, I am more convinced than ever that the Fragile X community will—in the not too distant future—be blessed with significant enhancements to clinical and pharmaceutical treatments that will profoundly improve the quality of life for our family members.

In closing I want to thank my fellow board members for providing me the opportunity to serve as a leader of this remarkable organization. I have deeply appreciated the support they have given me, especially from Board Vice President Brad Whitus. Thank you, Brad, for agreeing to serve as our next president. I can assure everyone we will be moving forward in terrific hands.

The author lives in Saint Mary’s City, Maryland with his wife Lisa, and sons Dillon, 17, with FXS, and 15-year-old Ryan, a carrier. Lisa’s extended family is impacted by FXS, FXPOI and FXTAS. Email: michael@fragilex.org
Moving Forward With Mike

My son Mike was 3 years old when he was diagnosed with fragile X syndrome. He is now 28. The literature I read in 1986 indicated that most boys with FXS were institutionalized during their teenage years. I must admit there were often tears when I thought of this possibility. So I am happy to report that I cannot believe what our situation is now compared to Mike’s younger years. I hope that telling our story will help parents of young boys so they might see light at the end of whatever tunnel they feel themselves in. Try to be positive—there is a lot of growth and development ahead. Many times I have felt Mike reached a plateau and that was where he would be the rest of his life, only to find he left that spot and moved forward.

One of the most vivid pictures I have of Mike as a young child is of him screaming. He has a sister and brother, seven and eight years older, respectively, and neither with Fragile X. We would attempt many activities typical in today’s world, such as eating at a restaurant, taking a trip to the mall, or attending a school activity. Most of these events were too much for Mike’s sensory system and he would begin screaming, which caused my husband or me to leave with him while the other parent stayed with the two older kids. We often experienced “the look” (as I termed it) from people no doubt wondering, “What are those parents doing to that kid?” Somehow we lived through those years. I felt my husband and I made it using the approach, “divided we stand,” meaning that one of us was with Mike and the other was with his siblings. Mike tests about a 55 IQ, and his speech development was much delayed, so I figure he used screaming to express how uncomfortable he was in so many situations.

In his early teens Mike had no interest in friendships, and he continued to find many social situations very uncomfortable. Although his screaming had stopped, he still was unable to express what was bothering him. He found relief from stressors by hiding in the boy’s bathroom or fleeing to the edge of a baseball field during team practice. When we were outdoors we had to keep our eye on him, because if we lapsed for a moment, he was gone and we would have to comb the area to locate him.

By high school Mike was 6-foot-3-inches tall and still needing to escape stressful situations. His long legs carried him away very quickly. During these years Mike’s stressors were no longer noise and chaotic environments. From observation I realized that particular individuals from his classroom or social groups were now the source of his stress. I think Mike had been present when these individuals had experienced a “meltdown,” or had a verbal disagreement with someone. Mike is very sensitive to emotional situations and they are very uncomfortable for him. Girls seemed to be a particular problem, maybe because they tend to be more emotional than boys. So Mike was usually getting away from a particular person, and interestingly, he never forgot those situations. If he saw that individual later, he would still need to get away from him or her.

We tried very hard to learn as much as we could about FXS so we could understand Mike better and provide an environment that supported him. I felt strongly that I wanted him to be as involved in life as he could handle, so I signed him up for many activities. I hoped that we could desensitize him to anxiety and stress and help him achieve some tolerance.

I have often said that special ed was never special enough for Mike. By that I mean teachers often did not understand the characteristics of FXS. So I kept handing teachers articles to read. In elementary school a teacher noted that Mike was spitting on the table and smearing it around. In actuality Mike had low motor tone and drooled. He was trying to clean it up and the teacher misinterpreted.
In middle school the reading content was way over Mike’s ability, so he became frustrated and acted out. High school brought challenges because there were 15 students in a small classroom, and Mike’s tactile defensiveness proved difficult to manage. I had several calls that Mike was erupting with foul language and often a literal knee-jerk reaction—with a size 13 shoe on his foot! Obviously Mike got in trouble for this behavior. At that point we arranged for several days a week of home schooling, which decreased his stress. Even at that stage he was unable to express what caused his “meltdown.” Either he was unable to mentally figure out what was happening or he did not have the language skill to express his discomfort. He was about 20 years old at the time, and I thought this was the best he would ever be. Thankfully, I was wrong.

Amazingly, the post-school years have been much better. I have an entirely different young man here, one I never expected to emerge. Just after Mike’s graduation from high school in 2005, we moved to Pennsylvania. Mike received state rehabilitation funding and the job hunt began. I found lots of activities for him to join, but I thought he would refuse to go. So it came as a surprise when I asked him if he wanted to join a bowling group and he said yes. Then I asked him if he wanted to play soccer and he said yes again. Never once did he want to hide out at home. Presently Mike is out four to five times a week at various social and sport events. I must admit at first Mike would locate himself at the back of the crowd, and it took two years before he felt comfortable with his new peer group. He worked three and a half years in a fast-food restaurant with good reports until a change in management occurred and Mike had a bad episode that caused him to lose the job. True to form, he was unable to relate what had happened.

While looking for another job Mike volunteered in a senior center serving lunches and cleaning up. It was a wonderful experience for him, as the personnel were very supportive and he was able to feel productive. He developed warm relationships with both the staff and residents. Though Mike got a job in a college dining hall earlier this year and is earning positive reports from managers and job coaches, he is welcomed back to work at the senior center during college breaks.

One tool we started using years ago is to set up a weekly calendar. Mike fills in his work schedule, YMCA exercise times, sport practice times, social events, doctor appointments, and chores. I feel strongly that this has been helpful for Mike in preparing him for the next week. Having him write out his schedule provides a way for him to accept each activity and agree to it. Whenever something pops up unexpectedly I verbally prepare him for it and most of the time he asks, “Not today?” To me this indicates that a sudden change of schedule is stressful for him. So I say, “Not today. In a couple of days.” Mike responds “Oh, O.K. Not today.”

At this point we have a very social young man who searches out friends in a large group and goes over to them to say hello and shake their hands. For a kid who could not stand noise and chaos, who had no friends, Mike has come a long, long way.

I do not want you to think everything is perfect. We still have occasional problems, but life is pretty good! At 28 years of age, Mike is still unable to relate his feelings to us and tell us if something is upsetting him. We continue to encourage it, though, and maybe that will be the next skill he achieves. I put a lot of effort into providing an environment in which Mike can be productive, social, happy, and non-stressed. I am ever vigilant in situations that are uncomfortable for him, and I try to figure out how to alter them. I am also trying to document this kind of information so that future caregivers will have a better understanding of Mike’s idiosyncrasies and be able to provide the best environment for him to continue on his forward path.

The author lives in West Chester, Pennsylvania with Mike and her husband Jim. Email: jimmariesclark@verizon.net
Readin’, Writin’ & Trick Shot Basketball

The fine motor skills required in handwriting were a challenge for Willie Gregory as he navigated the transition from relatively free-wheeling preschool life to a more structured typical kindergarten class last school year. It’s a rare kindergarten boy who can sit still long enough to churn out small nuanced markings with a pencil; rarer still when that boy has fragile X syndrome. Which just gave Willie a nice clear goal to shoot for as he, like many boys, re-upped for a second go at kindergarten this school year.

Willie, the youngest of the four children shared by Kate and John Gregory of Asheville, North Carolina, built a solid foundation his first kindergarten year, having been taken under the supportive wing of a small group of female classmates. “It was the greatest thing,” his mother reports. “They kept him on track with assignments, reminded him to get his pencil out, all kinds of things that he couldn’t yet manage for himself. The classwork moved pretty quickly, but I think they taught him more than any teacher or parent could have.”

This year, Willie transitioned to a different classroom with a different teacher, and his supportive cast had moved on to first grade. Such wholesale change is usually a very large pill for children with FXS to swallow. At least temporary backsliding is common and even expected. Willie, however, is thriving. “He loves to read and is a great memorizer,” says Kate. “Last spring he regularly recited several lines from Julius Caesar after watching his brother’s school Shakespeare recitation. But his most improved ability is in writing; he’s now figured out how to write down an answer to something, or to circle an answer on a piece of paper. That was impossible for him last year.”

Another change his mother notes: “He seems to be more patient. He can follow a schedule now and make his needs known, even if he’s feeling stressed. Last year he’d fall on the floor crying when things got too much, but this year he’s much more able to say, ‘I’ve had enough, I’ve got to leave now.’”

Like many parents of children with FXS, Kate had occasion to visit Willie’s classroom the first month of school when he wasn’t there in order to introduce his classmates to fragile X syndrome. The result, as in most all other such instances, was increased acceptance and empathy from Willie’s peers and teachers. “They like him and they see he works hard,” she says. “Having a kid with Fragile X is a huge stretch for teachers, but they have just scooped him up; everyone wants him to succeed. We know it’s not easy, and we’re taking it year by year. So far, though, so good.”

Willie enjoying the action at the home of his beloved Duke University Blue Devils basketball team.
Meanwhile, Willie amuses himself and continues to develop his motor dexterity by learning to play the drums ("He's not just banging," Kate reports) and indulging his passion in trick basketball shots, having been inspired by his beloved Duke Blue Devils and their trick shot master, Kyle Singler. (Note: see youtu.be/g85Qf0wWiKw for an unbelievable and fun two minutes. Then see Willie’s version at: youtu.be/sAxG-J6vFus.

“We have three basketball goals of different heights in the driveway and basement, and Willie shoots and practices most every day, even before school,” says Kate. “It’s one of the places he shines among his peers. He can easily make at least seven in a row and often beats his sister at HORSE. He’s a huge Duke fan and knows all the players. We let him watch Duke highlights as a reward, and we joke that he could probably learn calculus as long as we said Duke highlights were at the end of it.”

The author has edited the Foundation Quarterly since 2005.
Email: editor@fragilex.org

**Take a Foto for the Foundation!**

This Foundation Quarterly you hold in your hand is the product of many peoples’ efforts—not the least of them parents and friends who share digital photographs of their children and loved ones with us on a regular basis. There is no staff photographer at this journal—most all the photographs you see on these pages are due to the kindness and talent of amateurs with the desire to share their work with us. We invite you to be among them. In order to help you along, we’ve pulled together some basic guidelines to assist in your efforts. Please review them and then snap away for the benefit of interested and appreciative members of the Fragile X community. Photos are always welcome via email to: editor@fragilex.org.

1. **The most critical point and the chief determinant of whether your photo will be suitable for publication:** Please set your camera on the largest image size and the highest quality resolution it will allow. Different cameras have different terms, but the setting will usually be noted as “High,” “Super Fine,” “Ultra-High” or a similar term. Consult your manual or a knowledgeable friend or relative if you can’t find where you manage this setting.

2. Your final image should ideally be at “300 dpi” (dots per inch), which will make for a sharp (rather than blurry and faded-looking) image. To find dots per inch, click on the photo on your computer’s pull-down menu for something like “Inspector,” “Get Info” or “Document Properties.” DPI, along with image size and file size, should be noted there. Please note: the dpi cannot simply be typed in and changed (for instance, from 72 to 300 dpi) in your photo editing software; the image must be shot at a high dpi.

3. Most photos of adequate dimensions and resolution will be in a range from 500K to 2MBs on your file. If it’s under 100K, there’s little chance the photo will hold up for print media (though they may well be fine for the NFXF website; electronic media requires much lower resolution than does print).

4. Unless you are very adept, do not crop, resize or try to edit your image in any way. We would much prefer to worry about that.

5. Try to save your image in JPEG, PDF or TIF formats for easy transmission. Do not use heavy JPEG compression.

6. Next issue, we’ll focus on what to look for in composing and taking the photographs themselves.
Some of you may be searching for the “Focus on FUNdraisers” column, but from this point forward we will be recognizing our small event fundraisers in our eNews articles and on our NFXF Facebook page. We have found that this is the best way to provide the most up-to-date information about events as they happen.

In this new “Xpressing Gratitude” column, we will be recognizing many of our individual donors for their generous support of the foundation. Sincere thanks go out to all of our donors—as is all too obvious but bears repeating again and again: we couldn’t continue this critical work on behalf of the Fragile X community without you!

FX Donors Honor Roll

It gives us great pleasure to recognize the following donors for their gifts in the course of the last year (10/15/2010 through 10/14/2011).

$10,000+
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We would also like to recognize our Trustees, a special designation for those who have pledged financial support of the foundation at $1,200, $2,400, $3,600 or more annually for three years.

$3,600+ Annually
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Debra and George White

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Shorta Yuasa, MD
Jerome Zeldis

*In addition to personal donations, these people organized events that raised over $10,000 for the NFXF.

Think Membership!

Your annual membership with the National Fragile X Foundation helps spread the word about Fragile X while also helping the foundation provide free information and materials, referrals to support groups, and support for research. All members receive four issues annually of this very journal you are holding in your hand, courtesy email reminders of upcoming NFXF events and activities, and our email newsletter Fragile Xtras, which contains up-to-the-minute details on the latest happenings in the Fragile X community. To learn more, visit membership.fragilex.org or call (800) 688-8765.

The author is the development coordinator for the NFXF. For more about organizing or supporting fundraising activities, contact her at meghan@fragilex.org.
The National Fragile X Foundation engages a wide variety of funding sources in order to continue serving the Fragile X community in the multiple ways that it does. But at the very heart of our fundraising activities is our Annual Fund, always at year’s end, always an opportunity to reflect on what we have accomplished on our members’ behalf and what still beckons us forward.

Most of you reading this have probably already received our Annual Fund mailer, highlighted by brief stories of individuals and families who continue to move forward in the face of Fragile X. If you have not already done so, please use the remit envelope to send in your tax-deductible contribution, or log on to annual-fund.fragilex.org and click on the donate button.

It’s an exciting time in the Fragile X field, with tremendous forward momentum in research, treatment, therapy and education. Our Annual Fund allows all of us to make a collective statement that continuing on that forward path is vital, that the work the NFXF is doing to support the extended Fragile X community in its efforts to make the world better for those affected by Fragile X is worthy of our time and treasure.

Please give generously. THANK YOU!
The 13th International Fragile X Conference is designed to provide a forum in which professionals from the Fragile X and intellectual disabilities fields 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 all Fragile X -associated Disorders (FXD), which include fragile X syndrome (FXS), fragile X-associated tremor/ataxia syndrome (FXTAS) and fragile X-associated primary ovarian insufficiency (FXPOI).

Submitted abstracts will be accepted online at conference.fragilex.org/miami2012/call-for-abstracts/

Specific workshops will cover a broad range of disciplines and will be directed toward an audience of families, physicians, scientists, clinicians, therapists and educators. Presentation topics for families and clinical professionals will include medical treatments, speech, language and occupational therapies, educational techniques, opportunities for self-determination and independence, genetic counseling strategies and mental health interventions throughout the lifespan.

Based on the ongoing development of new therapies, the conference will include a focus on both pharmacological and non-pharmacological interventions (current and anticipated), evaluation tools, and evolving screening and diagnostic technologies. The conference will also highlight basic science presentations on functions of FMRP, molecular and cellular mechanisms in FXS, FXTAS, and FXPOI, as well as studies in animal models of FXD.

The conference will include a wide variety of other subjects including alternative interventions, public policy, advocacy and parent-to-parent support.
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