



The National **Fragile X** Foundation

# quarterly

WINTER 2002

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## What Is Fragile X Syndrome?

Fragile X syndrome is a hereditary condition which can cause learning problems in both males and females. It is the most common cause of genetically inherited mental impairment. The spectrum of intellectual involvement ranges from subtle learning disabilities and a normal IQ, to severe mental retardation and autism. In addition to mental impairment, Fragile X syndrome is characterized by a group of symptoms, which include physical and behavioral characteristics and speech and language delay.

Fragile X syndrome can be passed on in a family by individuals with no sign of the condition. In some families it is a problem that has been occurring for decades, affecting numerous family members through the generations, while in others, it seems to have caused problems in only one person. The National Fragile X Foundation has been helping individuals with fragile X, their families, and the professionals who work with them, since 1984.

Cover Art Logo by Andy Bobrow and Kathy Nagler

The Foundation Quarterly is published four times per year by the National Fragile X Foundation and is distributed to members and others by request. Please contact the Foundation regarding matters pertaining to the content or distribution.

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### Services offered:

Telephone consultation—*free*  
Basic informational packet—*free*  
Educational resources (books, audiotape, 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 newsletter—*fee*

### Services offered to:

individuals, families, professionals, institutions, and students  
involved with or impacted by fragile X syndrome

**Service Referral:** self or professional

**Eligibility:** all

**Service Area:** national and international

**Full Membership:** 2000

**Annual Contacts:** 20,000

**Website Visits:** over 100,000 annually

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

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

**Funding:** individual contributions

## Special Olympics - Family of the Year

# Florida Special Olympics Selects Family Affected by Fragile X!

To say that the story of Jonathan Doring is a “work in progress” would be a tremendous understatement. At just 21 years old, this young man has surpassed the limits set for him in his youth by doctors, he has challenged the world’s notions of what a person affected by Fragile X can achieve and has led a family on a quest to improve the lives of countless children and adults just like him.

The Doring family of Boynton Beach, Florida became actively involved with community service work when their son, Jonathan expressed a desire to play baseball like his older brother Mark. A few years earlier, his parents Kathy and Mark, Sr., had signed him up for Tee Ball Little League but it just wasn’t the right fit for Jonathan. Kathy heard of a new division of Little League called “Challenger.” It was designed specifically for children with mental and physical disabilities. Their involvement with Challenger would quickly grow over the years and become a family affair with Kathy organizing the league, Mark, Sr. coaching and Mark, Jr. playing as a “buddy.”

Looking to expand Jonathan’s competition opportunities, the Dorings became involved with Special Olympics in 1988 when Jonathan turned eight years old and became eligible to compete. Jonathan’s first Special Olympics sport: roller skating. Jonathan still participates in the speed skating and relay events in roller skating and has added bowling, tennis, softball and golf to his sports repertoire, winning numerous medals and ribbons over the years.

In addition to sports, Jonathan became a Special Olympics Palm Beach County spokesperson. County coordinator Rich Fleming says “this is a great accomplishment for Jonathan, since only three years ago he didn’t carry on conversations with most people and often chose not to speak at all.”



Jonathan Doring working hard as an official Sargent Shriver Global Messenger for Special Olympics Florida.

In 2000, Jonathan underwent an intense three-day training seminar to become an official Sargent Shriver Global Messenger for Special Olympics Florida. As a Global Messenger, Jonathan visits schools, civic groups and businesses for the purpose of recruiting new ath-

*Continued, page 4*

## Special Olympics - Family of the Year

*Continued from page 3*

letes, coaches and volunteers and educating the public about the abilities of people with mental retardation. Since that time, Jonathan has said the athlete's oath at several competitions and speaks at fundraisers on behalf of Special Olympics. Jonathan is also an Athlete Leadership Program member and has run in the Florida Law Enforcement Torch Run.

Jonathan's youth was spent attending elementary school at the JC Mitchell Special Education Center in Boca Raton. He was then promoted to Christa McAuliffe Middle School in Boynton Beach where he was gradually mainstreamed into many regular education classes. Jonathan proceeded to make the honor roll a record sixteen times and was inducted into the National Junior Honor Society.

Upon graduation from middle school, Jonathan attended Santaluces High School for the next five years. While there, he passed all of the classes required to receive a special education high school diploma and in May of 1997, Jonathan took the High School Competency Test to attain a regular education diploma. He passed the Math section but not English. Over the course of the next three years, he would take the English portion of the test eight more times. Finally, on December 15, 2000, in a special ceremony attended by the school district's Interim Superintendent, the Area Superintendent, the school Principal, many of his former teachers and his family, Jonathan was awarded his high school diploma.

Jonathan has since been awarded two college scholarships and is a student at Palm Beach Community College. He also works as an intern with RailAmerica, the largest short line railroad operator in the world and maintains a part-time job with Publix Supermarkets.

His most recent achievement was being selected to be an Olympic torchbearer for the 2002 Winter Olympics in Salt Lake City. In July, Jonathan will travel to the National Fragile X Foundation International Conference in Chicago to share his experiences with Fragile X and Special Olympics.

Mark, Jonathan's brother, is currently a college student at the University of Florida and has supported Jonathan all along. When he was younger, he often practiced with Jonathan and today, he attends his games to cheer him on. Mark, Sr., Jonathan's father, coaches bowling and has chaperoned at numerous state games. Kathy, Jonathan's mother, is a renowned organizer and self-starter. Kathy helped to initiate a Special Olympics tennis program at Delray Beach tennis center and in 2001, found a coach and site for Palm Beach County's new golf program. Kathy helps out wherever needed at local games, frequently volunteers at state competitions and serves on the Special Olympics Florida Board of Directors, the state public relations committee and serves on Florida's state families committee.

The Doring family has been instrumental in developing Palm Beach County's family program with Kathy and Mark serving as the family committee chairs for four years. In that time, they have produced many family events such as Palm Beach's family dinners, family mini-golf nights, family bowling nights and assisted with Palm Beach's biggest family event: their annual family picnic, which was attended by over 300 people last year.

The Dorings have all given much of themselves to make sure there are programs and services for people with mental retardation in their community. This was but one reason why Special Olympics Florida named the Dorings the 2001 Family of the Year.

In many ways, the Doring family's story is not unlike the stories of thousands of other families whose lives have been affected by Fragile X. What is remarkable about Jonathan is that he has, through the support of his family and community, continually shattered the stereotypes and myths about what he and people with disabilities "can't do" by showing the world his abilities, not his disabilities.

Amie Schank  
VP, Public Relations  
Special Olympics Florida

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## From the Executive Director

Robert Miller

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### Fundraising

Thank you to all of you who have responded to our holiday fundraising campaign and to those of you who are still planning on sending in your remittance envelope. We couldn't do it without you. In addition to the many, many responses to our holiday appeal, we continue to benefit from the efforts of individuals and groups around the country. While space makes it impossible to acknowledge every contribution, I do want to take special note of substantial support from the Swig Foundation, William & Deborah Parker and their 2nd Annual Champions for Children Celebrity Golf Tournament, The Fragile X Alliance of Ohio, and The Fragile X Resource Center of Missouri.

### Our Website

Have you seen some of the new features at [www.FragileX.org](http://www.FragileX.org)? Our home page has special icons for the newly diagnosed family and for the doctor. These pop-up windows provide quick, essential information, as well as links to more detail. We've also added a great way for you to review past newsletters. Just visit our "Newsletters" page and let your cursor hover over the cover of any issue. The table of contents will then "magically" appear! If you're interested in non-nonsense commentary on some of the latest research, check out our new "Research" area. And, lastly, we now have selected NFXF articles in Spanish on our new "Espanol" page.

Speaking of the website, on December 3, 2001, Dr. Dale Fast, Associate Professor in the Department of Biology at Saint

Xavier University, Chicago, was presented with the Saint Xavier Award. This is an award given annually to one faculty member, who has shown an outstanding record of service to the University and community. Dale's long list of service activities spanned the last 30 years, from his time as a Peace Corps volunteer in Africa to his most recent service, the extensive revision of the National Fragile X Foundation website, completed with Dr. Gail Harris-Schmidt during his sabbatical leave in Fall, 2000. A letter from Robert Miller, Foundation Director, was read as part of the ceremony honoring Dale. An article in the Saint Xavier University faculty newsletter also included an article on fragile X and the website. Congratulations to Dale!

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## Prevalence of Fragile X Revisited and Revised

IN THE WINTER 2000 FOUNDATION QUARTERLY, Dr Randi Hagerman addressed the question of the prevalence of fragile X syndrome across the entire population. In our editor's note, we affirmed our commitment to review new data and adjust our published statistics as appropriate. Based on a recent report from the U.S. Center for Disease Control (CDC) we here revise our prevalence statement. Our year 2000 article cited prevalence of the fragile X, full mutation in boys as: 1:2000 and in girls as: 1:4000. These numbers were meant to include not only those with mental retardation, but also those with learning disabilities and emotional problems without mental retardation.

Based on the recent CDC report, which is itself a review of existing prevalence studies, the National Fragile X Foundation will henceforth publish a prevalence for males, with the full mutation, of 1:3600. The CDC did review *some* studies which did include *some* individuals with learning disabilities in addition to those with mental retardation.

Prevalence in females is less well understood and certain because of a lack of studies. However, it is likely that at least 1:4000 to 1:6000 females has the full mutation for fragile X—a figure higher (more prevalent) than the CDC estimate. We propose this adjustment because girls with fragile X often present with more mild involvement than boys, including learning disabilities and emotional problems, and thus may not be screened or tested.

Whatever the prevalence of fragile X, it remains the most common known cause of inherited mental impairment. Some of the uncertainties and differences of opinion stem from differing definitions of just who is "affected." Whether just those with the full mutation are affected, and to what degree those diagnosed with learning disabilities and/or emotional problems have fragile X, are two such questions which can impact prevalence. Differences in defining the cut-off point between pre-mutation carriers and those with a full mutation also cloud the issue. Most importantly, a lack of large-scale, comprehensive studies allows for wide-ranging differences of opinion. While a greater degree of certainty regarding prevalence would be desirable, and important in the quest for increased government-funded research, what is most important is that fragile X syndrome:

- is prevalent,
- has a significant impact on society,
- is relatively unknown,
- is under diagnosed or largely undiagnosed,
- is a key to understanding other neurodevelopmental disorders such as autism, and,
- as such, needs our full attention—
- no matter how prevalent.

**To view the CDC report, visit:**

[http://www.cdc.gov/genetics/hugenet/factsheets/FS\\_FragileX.htm](http://www.cdc.gov/genetics/hugenet/factsheets/FS_FragileX.htm).

## UCLA Studies How Boys Respond To Sound

Elisabeth Dykens, Ph.D. and Edward Ornitz, M.D. of UCLA invite you to participate in a study of how boys with fragile X syndrome respond to sound. We are particularly interested in how the response to a quiet sound affects the response to a loud sound when a quiet sound precedes a louder sound. Why is this important in fragile X syndrome? Based on studies with fragile X mice, it looks as if these mice are responding differently than we would expect to soft sounds, and also differently than we would expect to louder sounds. It is important to see if this same unusual response pattern to sound is also seen in humans, and how it may relate to the sensory and behavioral difficulties seen in many boys with fragile X syndrome. If the patterns to sounds are similar in both mice and people, then ultimately we can use the mice models to evaluate potential pharmacological or gene therapy treatment that hold the most promise for boys with fragile X syndrome.

What do we specifically need from you? We would like to work with boys with fragile X syndrome aged 8 through 17 years and their parents for about three hours. We would assess the boys' responses to sounds and administer a brief problem-solving task. Equally important, we would also ask parents about their son's everyday adaptive skills and performance, and any negative behaviors that might get in the way of these skills. In return, we would give parents individualized written feedback about their son's performance, as well as a token \$25 stipend and reimbursement for your mileage to and from UCLA.

If you are interested in participating or have any questions about this study, please call the project coordinator Dr. Beth Rosner at 310-794-9201. Thank you in advance as we work together to improve the quality of life for persons with fragile X syndrome and their families.

Announcing the 3rd in a Series  
of Free, NFXF Sponsored

## Online - Webcasts

### Question & Answer Sessions with Fragile X Specialists

Please join Marcia Braden, Ph.D.,  
**ONLINE**

**Tuesday, March 5, 2002**

**5:00PM Pacific**

**6:00PM Mountain**

**7:00PM Central**

**8:00PM Eastern**

Dr. Braden is a licensed psychologist in private practice in Colorado Springs, Colorado. She specializes in the learning and behavior problems of individuals with fragile X. Dr. Braden also works with individuals, families, school districts and hospitals in designing curricular and behavioral models. She has written numerous articles on fragile X syndrome as well as developed educational materials. She is the author of *Fragile, Handle with Care, Understanding Fragile X Syndrome* and a recent revision entitled *Fragile, Handle with Care, More About Fragile X Syndrome Adolescents and Adults*. She has published a Curriculum Guide, LOGO Reading, Educational Training Tapes, co-authored a chapter in *Fragile X Syndrome, Diagnosis, Treatment and Research*, edited by Hagerman and Cronister, Johns Hopkins University Press and *The Fragile X Child*, Woodbine House. Dr. Braden is a member of the National Fragile X Foundation's Scientific & Clinical Advisory Committee.

**To participate in the online session, visit  
[www.FragileX.org](http://www.FragileX.org)**

Click on the "LIVE Q&A Online" icon. Participants will be able to phone-in questions, email questions, or simply listen to Dr. Braden on their computers. (Internet connection and computer speakers required.)

# Chicago Prepares For Your Visit

By Dr. Elizabeth Berry-Kravis

*EDITOR'S NOTE: Dr. Berry-Kravis is a Pediatric Neurologist based at Rush-Presbyterian—St. Luke's Medical Center in Chicago*

As someone with many years of experience treating children and adults with fragile X, I am pleased to tell you about some of the exciting presentations being planned for the National Fragile X Foundation's 8th International Fragile X Conference. The conference is being hosted by the dedicated families that make up the Fragile X Resource Group of Greater Chicago, who are hard at work on this major project. The conference will incorporate new topics of interest to both parents and professionals. Family-focused and professional-focused presentations will again run concurrently but with an even greater emphasis placed on serving the needs of each target audience.

There will be an increased number of sessions focusing on adults, with topics including aggression, financial planning, and legal issues. In addition, because questions from parents are often not specifically answered at scientific sessions, popular workshops like those on psychopharmacology and neuropsychology will be divided into research-focused presentations and parent-focused presentations.

Parent presentations will also include: *understanding and using psychological test results to obtain specific interven-*

*tions at school; psychopharmacology and the benefits, pitfalls and specific symptoms different medicines can target; insurance advocacy; camp programs; social skills training; math interventions; visual problems; and many more topics of importance to families.*

There will be an extensive array of educational presentations covering inclusion, curricula, and implementation of best practices for students with

fragile X. Sessions on educational programming and methods will be concentrated over a two-day period so educational professionals can access this information without attending the entire meeting.

Recognizing that in the last two years we have learned much about what FMRP does in the brain and are moving

ever closer to identifying the specific neurochemical mechanisms which are altered in the absence of FMRP, this meeting will include significantly more neurobiology and brain-based research than previous meetings. (Some of these findings are already leading toward the development of new drugs targeted at brain mechanisms in fragile X syndrome. Although these new classes of medicines are not yet available for trials in children, early trials in adults are now being planned. These new medicines may work much better than traditional drugs that treat symptoms rather than the underlying mechanism, and

may in fact work on cognition rather than just behavior. These potential new treatments will be discussed including a presentation on an exciting collaboration involving the RUSH-Presbyterian-St. Luke's Medical Center Fragile X Clinic, the Fragile X Clinic at the MIND Institute at UC Davis and Cortex Pharmaceuticals, Inc. who will soon begin a trial of AMPAKINE® CX516, in adults with fragile X syndrome and autism.)

Other "hot" research topics will include tremor and neurological problems in male premutation carriers from the collaborative project at RUSH in Chicago, UC Davis M.I.N.D. Institute, University of Colorado, Children's Hospital, Denver, and a review of current reproductive options and programs for pre-implantation diagnosis.

In keeping with the rapid pace of progress in research and therapeutic options for patients with fragile X, the 8th International Fragile X Conference promises to be a very exciting meeting with plenty of new information for both parents and professionals.

Together with the members of the Fragile X Resource Group of Greater Chicago I look forward to sharing our beautiful city with you. Please join us July 17-21, 2002 for an event you will never forget.

## REGISTER TODAY!

For International Fragile X Conference registration

please turn to pages 18-19

**Fragile X Resource Group of Greater Chicago**  
New Phone: 847-465-1245  
New Fax: 773-779-9061  
New Email: fragileXchicago@aol.com  
New Web: <http://www.fragileXchicago.org>

# Adulthood and Fragile X Syndrome:

**Andrew Levitas, M.D.**

*Medical Director, Division of Prevention and Treatment of Developmental Disorders,*

University of Medicine & Dentistry of New Jersey/SOM

WHEN THINKING ABOUT the challenges faced by persons with fragile X syndrome in adulthood, and their families, it is important to consider both the issues they share with other persons with developmental disabilities and those particular to fragile X syndrome. Those particular to fragile X syndrome may some day find answers in laboratory and clinical research. Those shared with other persons with developmental disabilities are issues of social policy and ideology, and are more likely, at least in the short run, to find answers in social and political arenas.

The perspective of mental health is a view of the maintenance of coping skills, the continuity of daily life, and hopefulness for the future necessary to emotional stability and fulfillment. Dare one say happiness? A diagnosed Psychiatric Disorder ordinarily plays only a part; more important are the family and community systems with which a person is involved and by which he or she is supported. Conflicts within these systems do not imply a psychiatric disorder, but can give rise to problems of mental health including: loss of coping ability, loss of connectedness to daily life, and loss of hope in the future. This loss is experienced as “stress” (more properly, anxiety and/or chronic sadness or anger) and can, in vulnerable individuals, lead to true psychiatric disorder. The goal of mental health care is to prevent this loss.

I write the following as a child and adult psychiatrist with 25 years’ experience in the field of developmental disabilities, and also as a family member of a person with developmental disabilities. I think my biases will be plain. I do not expect them to be shared universally, and hope these observations will spark lively reader response. I think it is possible to set out observations of the current challenges, and not possible to suggest

solutions applicable, comfortable, or even possible, for *all* families in *all* times and places. It is also worthwhile for the reader—and the writer—to remember that a psychiatrist sees the problems of the people for whom the system is not working. I am therefore more aware of its shortcomings than of its successes.

The process of choosing among residential and vocational programs for an adult with fragile X syndrome and developmental disabilities is in some ways similar to choosing a college. There is a huge array of possible choices, with many different and sometimes conflicting goals to weigh.

## Residential and Vocational Alternatives

Public Law 94-142 mandates educational alternatives for all children and adolescents with developmental disabilities through age 23. After that, each state provides an array of residential and vocational programs. These, not unlike our college system, vary not only by state but by localities within each state. They may be public, private subcontractors, private non-profit or for-profit. Almost all have a basis of funding in Medicaid or

Medicare/Medicaid, and are more or less subject to those guidelines. What this means in practice is that treatment planning, staffing patterns, and other requirements are driven by the standards of the dominant funding agencies, in a way analogous to the way local school districts vary (but perhaps not much) in their approaches to following state guidelines. Just as educa-

tional planning in the schools follows the Individualized Educational Plan (IEP) format, treatment planning for adults with developmental disabilities follows the Individualized Habilitation Plan (IHP) format, and the two closely resemble each other in their patterns of periodic goal-setting meetings bringing together family members and/or guardians, case managers and agency professionals and caregivers. As in the IEP, this is the place where individual needs are negotiated and reconciled with the available services.

The range of services varies from provider agency to agency. For example, residential services may range from independent apartments with “follow-along” counseling,

through supervised apartments to group homes, with some agencies providing the full range of services and some not. Similarly, vocational programs might range from competitive employment with job coaching services through supervised crew labor and sheltered workshop employment to adult day care. Some agencies provide both residential and vocational/day care services, others only one (meaning the person and family must deal with two sets of providers). Securing some needed services may mean shifting from one agency to another, with all the attendant trade-offs if different agencies offer different but equally desirable services, a process of varying difficulty.

The guiding values—the ideology, if you will—of the system is Normalization, and the principle of the Least Restrictive Environment. That is, every person with developmental disabilities should be in a situation that is as close as possible to what is “normal” for a person of that age and in a residential and vocational environment that least restricts his or her freedom. As principle this is *laudatory*. In practice it suffers from, first, the inescapable fact that what is “normal” for one person is not “normal” for the next, and, second, that the “least restrictive environment” is in fact to be chosen from the limited list above. Attempts to define “normal” in my experience have led to instances of persons with severe developmental disabilities being required to give up their beloved World Wrestling Federation and Disney posters in favor of “more age appropriate” (in someone’s opinion) room decoration, to cite a relatively trivial example.

From a mental health point of view, what is “normal” is the right to choose one’s room decoration, not the nature of what is chosen. A “normalized” environment may not, in fact, be the “least restrictive” if someone else is choosing what is considered “normal.” Attempts to force “normal” choices on persons who already have pronounced likes and dislikes has been the source of quite a number of psychiatric evaluations for me over the years. Imagine living in a dormitory that required everyone to hang only art museum posters on the walls.

“Least Restrictive Environment” also has come to mean moving each person along to his or her maximum level of independent functioning. This, too, is a laudable goal, but

**The perspective of mental health is a view of the maintenance of coping skills, the continuity of daily life, and hopefulness for the future necessary to emotional stability and fulfillment. Dare one say happiness?**

# A Mental Health Perspective

one which in practice means meeting goals set by others. A person with all the abilities needed to hold a competitive job, for example washing dishes at a fast food restaurant, might be reluctant to leave his friends at a sheltered workshop or crew labor to be the only person with developmental disabilities at the local burger place, or might discover this isolation only after making the change. On the other hand, the person with all the skills necessary to work at crew labor, but with a history of “behavior problems,” may have problems indeed when he must “first prove himself” at a sheltered workshop. A person may be quite independent in the skills of self-care and daily living, more than able to live in an independent apartment, but not have the same level of skills and independence at work, requiring a more flexible approach to residential and vocational needs than some programs are capable of. In the case of persons with more limited cognitive functioning, or with limited tolerance for change and uncertainty, a more structured environment, which looks to an observer to be “more restricted” actually permits the person more comfortable functioning and therefore access to more activities than he or she could tolerate in a less structured setting.

All of these are examples of situations that have led to psychiatric consultation, illustrating if nothing else, that as it is for all of us, the most common source of stress in life is the inflexibilities in our environment. *All such situations are potentially capable of being resolved within the IHP process (the “I” in IHP is supposed to stand for “Individualized”), but this requires active intervention and advocacy.*

Transition to a residential program should ideally be gradual, analogous to how all of us spent longer and longer times away from home until we were on our own. In fact, residential programs vary widely in their approach to new residents. One method of transition families may find helpful is to seek respite services at a residential agency before the individual moves in. This affords new residents and their families the opportunity to both test the environment and meet other residents and caregiving staff. This does not, however, address the issue of roommates. Few residential programs offer single rooms—imagine college freshman year with no option to switch roommates. Spending advance time

with prospective roommates is desirable where possible.

Caregivers at residential and vocational programs are commonly paid minimum wage, and may compensate by working many shifts. Often these jobs are taken by students saving for the next step in their educations. The result is a high rate of staff turnover, which can be hard on persons with developmental disabilities for whom relationships with caregivers are so important, and who may tolerate change poorly. Short of a major change in social policy there seems no solution in sight for this. One cushion built into the system is that there are always multiple caregivers, minimizing the chances that all of them will leave at the same time. The ultimate “cushion” of course, where possible, is ongoing family involvement, and friendships. Often friendships between children with developmental disabilities is not encouraged, in the belief that they have nothing to teach each other (as if life were one long therapy), and this is poor preparation for adult friendships. Both residential and vocational programs could do more to promote shared activities. Often local developmental disabilities organizations offer recreational programs (beyond Special Olympics, which also in my view does less than it could to promote relationships between persons with developmental disabilities) including dances and trips.

What humanizes and softens, what sounds—*when one focuses on its shortcomings*—like a harsh system is the presence of many dedicated and caring people who are aware of these shortcomings and work with people and families to smooth the sharp edges. Every group home and every work crew is a community, a small town with constantly shifting circumstances, again not unlike a college with a new outgoing and incoming class and faculty every year. It is the relationships within that community that create the experience of it, meaning a constant “dance” or series of negotiations as life goes on.

## **Psychiatric Disorders common in adults with fragile X syndrome**

We do not understand enough about the normal course of changes in brain chemistry to know why many psychiatric disorders have their onset in early or mid-adulthood. The trajectory of these changes in persons with fragile X syndrome is a further unknown. We

do know that some of the psychiatric symptoms and disorders common in children with fragile X syndrome continue into adulthood, and may evolve. Anxiety disorders, including Panic Disorder, are known. Obsessive-Compulsive Disorder (OCD) is known to begin or intensify. Mood instability may not worsen, but, in an environment requiring more flexibility than a person with fragile X syndrome is capable of, may become a more frequent issue. Mood disorders are also seen in persons with fragile X syndrome. Seizures, which may have their onset as late as the early 20’s, may increase the risk of any or all of these problems. Autism spectrum disorders, common in fragile X syndrome, may be associated with more severe versions of all of these.

Fortunately, medication interventions are available for all of these problems, and (not surprisingly, since they are tested in adults) can be more reliable and predictable in their actions than they are in childhood, and their side effects better tolerated. Behavioral intervention, which is also very helpful, varies widely in availability from agency to agency, as does availability of psychiatric consultation. Many agencies depend upon the public mental health system, which also varies widely in its expertise with persons with developmental disabilities. It is most advisable always to seek psychiatric evaluation by someone experienced with persons with developmental disabilities, where available, and to supply any mental health personnel with information about fragile X syndrome.

## **Issues Especially Relevant to fragile X syndrome**

Evaluating the characteristics of the adult developmental disabilities system in light of the needs of persons with fragile X syndrome, it is possible to anticipate some of the issues likely to be encountered.

The familiarity of residential and vocational caregivers with fragile X syndrome varies widely, to the point where it would be unwise to assume it. Even where caregivers may have heard of fragile X syndrome, they may “know” only that “everyone with fragile X syndrome has Autism,” or not know that eye contact is unpleasant. They may never have met a family with more than one member with developmental disabilities, and understand the balancing of needs that this entails.

*Continued, page 10*

## Adulthood in Fragile X Syndrome

*Continued from page 9*

It is best to be prepared with reader-friendly literature and the National Fragile X Foundation web site. ([www.FragileX.org](http://www.FragileX.org)) Do not be surprised by caregivers and professionals who think this information is irrelevant, or who think that the needs of all persons with developmental disabilities are the same.

Shyness, poor tolerance of crowds, need for sameness and predictability, stereotypic movements and unusual speech may limit vocational options at any level of cognitive ability. This may unfairly lead to exclusion from competitive employment for someone perfectly capable; just as often it may lead to “promoting” a person with skills but unusual needs to a job he or she cannot tolerate.

Similarly, obsessive-compulsive needs (rituals, poor tolerance of change) may be poorly understood by residential caregivers, who may have no idea that these characteristics and behaviors are not under the individual’s control, and may lead to conflicts and frustration, even to poorly-conceived interventions to eliminate them. Here again, patiently informing caregivers and seeking expert consultation is the most advisable course, *again, if necessary, using the IHP system to negotiate and create appropriate goals and interventions.*

For persons with fragile X syndrome and autism spectrum disorders, residential and vocational programs specifically for persons with autism spectrum disorders may be a better fit than programs more broadly serving all persons with developmental disabilities. Such programs are more understanding of the poor tolerance of change, compulsions and rituals, and limited social engagement typical of persons with autism spectrum disorders, are less likely to have inappropriate expectations of the person, and are more likely to have appropriate interventions available when problems arise.

This brings up the issue of special residential programs for persons with fragile X syndrome. This has been tried for persons with Prader Willi Syndrome (PWS) because they require close supervision of food intake so restrictive that it would violate the rights of anyone else in the house who should have free

access to food. While this solves some very important problems— unrestricted food access for persons with PWS can lead to fatal health consequences— it does not guarantee that everyone in the house has a friendly roommate. Having the same developmental disabilities syndrome doesn’t guarantee friendship, any more than having high blood pressure does. However, so many people with fragile X syndrome have compatible temperaments and behavioral traits that, if one must room with a stranger, it could be worth trying to at least have a roommate with fragile X syndrome, or failing that, a roommate at home with sameness, obsessiveness, and shyness. This doesn’t necessarily mean someone who shares those traits, just someone who tolerates or enjoys

**...the likelihood of success in adulthood can be maximized by attention to family and community... the cultivation of friendships [and] the identification of dedicated and caring people knowledgeable in fragile X syndrome...**

them. *A fragile X syndrome residential program would be subject to the same problems of funding and staffing as any other residential program, but would offer the advantage of ensuring appropriate staff training.* There is also no reason short

videos on fragile X syndrome could not be viewed by any new caregiver in an existing program.

None of this is an argument for or against a fragile X syndrome residential program. It should be clear, however, what problems such a program would and would not solve.

Another approach to the issues of adults with developmental disabilities, fully applicable to persons with fragile X syndrome, is that of customized, family-constructed services. In one version, a family, assisted by a broker, assembles a program of services to be purchased and creates a budget to be submitted to their state Developmental Disabilities agency. This can include a family-supervised apartment, in-home supervision, or the like. The advantage to such an approach is that the family remains in control of daily living and day program, in effect creating its own IHP. Two families known to me have tried this, with mixed results. One created, with the participation of the person with developmental disabilities, a committee of experts— therapists already involved with her or known to her— with whom she meets regularly to discuss her progress and needs, in effect sharing

and distributing a parental role in her life. This has worked about as well as, say, sending a teenager off to a distant college— well for the most part, with a few alarming scrapes. A second family set up a system of in-home day care for a family member with severe developmental problems. After about a year, the mother described this as:

“Running a group home for one— I’m the CEO, personnel officer, and back-up for whoever doesn’t show up, and when they do show up they are in your home, part of your life, like a nanny, so you have to be very careful who you hire.”

This is not a solution suited to everyone, or available everywhere yet.

The chances are that none of the above are sufficiently specific to apply to any one person or family. They cannot be. The adult developmental disabilities system in the United States is far too varied for anything but the broadest of generalities. Current approaches to health care in this country do not in my opinion offer much hope that we will soon see a system that attempts to accommodate the individual rather than fit him or her into existing services, whatever the fit. The lives of persons with fragile X syndrome will remain dependent upon the vigilance of their families and guardians and their advocacy, much as during the school years.

There is obviously no one best way to approach these issues or solve these problems, no ideal program or all-encompassing collection of programs. If there were, everyone would be doing it. However, much like success during school years, the likelihood of success in adulthood can be maximized by attention to family and community systems, active intervention and advocacy, ongoing family involvement, the cultivation of friendships, the identification of dedicated and caring people willing to work with people and families, identifying and working with professionals knowledgeable or interested in fragile X syndrome, and the effective use of medication. All of this cannot of course guarantee success, but in whose life is success guaranteed? It is also therefore important to note that unless some important reality is being neglected, there can be no question of judgment of any one family’s approach. All any of us can ever do is the best we can.

## FRAXA Update

### 2001: A Breakthrough Year for Research

Michael Tranfaglia MD

Medical Director (and fragile X parent)

FRAXA supports biomedical research aimed at finding a specific treatment for fragile X. In 2001, progress accelerated to an astonishing degree.

It all started during last year's Banbury Conference, held each March at Cold Spring Harbor on Long Island, and sponsored by FRAXA and the National Institute of Mental Health. These conferences gather small groups of fragile X researchers to share results and spawn collaborations—and it has worked better than we could have hoped!

In November, the prominent journal *Cell* published papers co-authored by FRAXA grantees Robert and Jennifer Darnell and Banbury-attendee Steve Warren. The papers identified proteins which are regulated by the fragile X gene. One protein, MAP1B, stands out: its levels are significantly increased in fragile X. In *Cell*'s next issue, FRAXA grantee Kendall Broadie showed that deleting the fragile X gene in fruit flies causes cognitive problems. Those problems can be

reversed by also deleting a second gene—the fruit fly version of MAP1B.

The editors of *Cell* highlighted the importance of these papers by publishing an extensive commentary along with the articles. This work gives us an exciting new target for drug discovery, since excessive function of one protein appears to account for many fragile X symptoms—at least in fruit flies. The goal of follow-up studies will be to find drugs which can partially block the relevant functions of MAP1B, in a safe and selective way.

You can reach us at:

**FRAXA Research Foundation**

45 Pleasant St., Newburyport, MA 01950

Phone: (978) 462-1866, Email: [fraxa@mediaone.net](mailto:fraxa@mediaone.net)

Web: [www.fraxa.org](http://www.fraxa.org)

*Editor's Notes: The Fraxa Research Foundation is a 501 (c)(3) nonprofit corporation not affiliated with the National Fragile X Foundation. The opinions expressed in this update do not necessarily reflect the opinions of the NFXF and/or its Scientific and Clinical Advisory Committee.*

*For additional commentary on this research, visit the NFXF's website's Research page at [www.FragileX.org](http://www.FragileX.org)*

### Chicago Supporters Race to Support Fragile X Syndrome

By Gail Harris-Schmidt

On October 21, 2001, the Fragile X Resource Group of Greater Chicago sponsored a 5K race, children's games, and a silent auction to raise funds for the National Fragile X Foundation and to raise awareness about fragile X syndrome. The event was co-chaired by the Schmidt and Urycki families, and Lynda Canel designed the event invitations. The event raised \$10,500 for the Foundation and generated local publicity, including an interview with Sarah Urycki, mother of a son with fragile X syndrome, published in a local paper. A number of high school cross-country team members volunteered by gathering items for the silent auction, hanging posters in the community, staffing the registration tables, and running or timing the race.

There were over 70 racers, including families of children with fragile X, classmates, teachers, neighbors, high school cross-country team members, and local race enthusiasts, many of whom knew nothing about fragile X syndrome before the event. Dr. Elizabeth Berry-Kravitz, Director of the Fragile X Clinic at Rush-Presbyterian-St. Luke's Hospital won a first place ribbon for women in the 40–49 age range!

After the race, the park lodge was the site of a silent auction, children's games, and a picnic lunch. A local Boy Scout troop and many of their friends became involved and staffed the children's games, the Saint Xavier University Speech Club staffed the picnic lunch, and Avis Primack and Maureen Schmidtgall of the Resource group provided great help with the silent auction. We met families with children who have fragile X and who are new to the Resource Group, and became reacquainted with others who have been in touch by phone and email. One of the real satisfactions of the event was a sense of raising awareness about fragile X. We overheard a father explain fragile X syndrome to his son, who is a classmate of a boy with fragile X. One of the cross-country team members said by wearing his race t-shirt to school, he had received at least five questions a day about, "What is fragile X?". Donors of items for the auction and food for the racers and lunch guests all learned a little about fragile X syndrome. In addition to the funds raised, it was good to feel that a difference had been made in our community.



# Parent's Forum

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EDITOR'S NOTE: When I first read the article below, it really brought home how my own mother must feel now that she knows our family has been affected by fragile X. This great-grandmother writes from her heart when telling how her own family has been affected, and how they have all come together to overcome the challenges of fragile X syndrome. I can honestly say that this family is special! I had the pleasure of meeting Theresa, Wayne, Keith, Karen and little Jacob in 1999. What an inspiration they are to me and, hopefully, to many other families. I want to thank them for sharing their story.

- Cindi Rogers, [CIJRogers@aol.com](mailto:CIJRogers@aol.com)

## Fragile X: The Past, Present and Future As Seen Through the Eyes of a Mother, Grandmother and Great-Grandmother

By Theresa M. Houselog

I've often thought about sharing my experiences with fragile X syndrome. The timing was never just right. How easy it is to put off the important things in life. I believe that if my story helps one family struggling with the elements of fragile X, my time and efforts will be worthwhile.

### **The Past**

To begin, I would like to share some background and history of fragile X in my family. My maternal grandparents had 10 children: 6 daughters and 4 sons. (My Mother was one of the daughters.) None of the sons manifested any characteristics of fragile X syndrome. However, most all of the daughters manifested some indication of being either a carrier or an affected female. Thus, we have concluded that the genetic disorder is traced back to our grandfather, a Swiss immigrant.

I am the oldest of eight children: 5 girls and 3 boys. Five of the eight have been tested: 4 girls and 1 boy. Of those, three of the girls, including myself, and one boy tested positive as carriers. Two of my brothers have not been tested. My youngest brother has a grandson that tested positive and is affected, therefore, we knew he was a carrier. One nephew and two first cousins were diagnosed as fragile X affected males in 1993 after their caregivers pursued the notion that there were similarities in their history of physical and emotional development.

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My personal introduction to fragile X occurred eight years ago (1993). My daughter, Karen, called one evening crying. “Mom, Jacob has been diagnosed with fragile X syndrome” she said. My knowledge of fragile X was very limited. However, what I did know was that it was the second leading cause of mental retardation. I was devastated and cried along with Karen— for her, for Jacob and for myself. It was like being struck by a bolt of lightning. We were all aware of Jacob’s slow development. He was already in physical therapy for hypotonia at the suggestion of his pediatrician, who also suggested some neurological tests, including a test for fragile X syndrome. A meeting with a geneticist was scheduled. Karen and her husband, Keith, were given a lot of information and it was evident that there would be some serious decisions to be made in establishing a treatment plan for Jacob. So much was happening and so quickly. To add to the shock and concerns, Karen was pregnant with another boy. Her sister, Mary Kay, was also pregnant, but did not know the baby’s gender. Nor did we know if she was a carrier of fragile X. Karen’s obstetrician suggested she be tested. The results indicated that she too, was a carrier.

The rest of the immediate family was informed of Jacob’s diagnosis. My heart ached. I didn’t know where to go or what to do. I was reluctant to tell anyone, not friends, not extended family, not siblings. I didn’t know why, but I felt I had to learn more about this disorder before I could talk about it. I felt guilty, sad and anxious all at the same time. I did know that I had to support my children. There was no point in looking back, feeling guilty, or feeling sorry for myself. Determined to learn all we could about this phantom disorder, we decided to gather as much information about fragile X as we could, as quickly as we could, and from whatever source we could

find. We focused on being positive in our research for some answers to our many questions.

Karen gave birth to another son just weeks after Jacob was diagnosed. His blood was immediately sent to the Mayo Clinic for testing. Subsequently, the test came back negative— some good news for a change! Meanwhile, our daughter, Mary Kay, gave birth to a little girl. Blood was drawn at birth and sent to the Mayo Clinic. The blood was lost en route. Everything seemed to be going wrong. Our world was topsy-turvy. What do we do? Who could we turn to? I was overwhelmed. I prayed for guidance.

In June of 1999, my husband, Wayne, and I accompanied Karen, Keith, Jacob and Austin to Denver to participate in a fragile X family history research project at Children’s Hospital of Denver. Having been invited to participate in this research study, we were eager to learn and to share.

In the course of the testing, I was diagnosed with depression. I experienced anxiety and relief all at once. I had not been feeling like myself for the previous year and a half. I was always anxious, suffered from insomnia, was prone to panic attacks and had slowly withdrawn from family and friends. It was all I could do to get out of bed in the morning, face the day or talk to anyone. If I had to go to an event where there were more than a few people, I worried about it for days before. I realized I needed some professional help.

When we returned home, I scheduled an appointment with a psychiatrist. With the encouragement of my family, an understanding psychologist, and a good psychiatrist, I slowly made progress in treatment. I am still in maintenance and medication management, but doing well.

*Continued, page 14*

## Fragile X: The Past, Present and Future

*Continued from page 13*

On June 27, 2000, our great-grandson, Nicholas was born prematurely and with some difficulty. He was not breathing when he came into this world, and developed respiratory difficulties, which is not unusual for a premature baby. Since the doctors were drawing blood for other medical reasons, it was suggested by the family to have him tested for fragile X. The test came back positive—he was affected. Little Nicholas has had, and continues to have, numerous medical problems, some of which are related to fragile X and some not.

By this point in time we knew for certain that of our six children, our oldest son, two youngest daughters and oldest granddaughter were fragile X carriers. One grandson and one great-grandson were also affected. Karen's daughter has not been re-tested since the loss of her blood. All of the immediate family has been tested with the exception of a nine year-old grandson and 6 year-old granddaughter. They exhibit no physical or developmental characteristics of fragile X, but we know they are at risk of being a carrier.

### **The Present**

Our granddaughter, Nicole (Nicholas' Mom) is an extraordinary young single mother. She works, attends college, and is dedicated to whatever it takes to help her little son. Jacob's parents are also extraordinary parents. Jacob is now 8 years old and is non-verbal, but has outstanding comprehension. He communicates through sign language and computer technology. He is attending an inclusive school where he is amazing even his teachers.

Great-grandparents, grandparents, parent(s), aunts, uncles and cousins have formed a circle of love and support for Jacob and Nicholas. They light up our lives and play very special roles in our family. In the midst of all of our trials and tribulations of the past 9 years, our family has become stronger and even closer. We all have a universal goal; to love and support each other in good times and in difficult times. Yes, there is still a long road ahead, but somehow it doesn't seem quite so bumpy.

Every now and then, when I'm forced to face a situation that seems overwhelming, I think "Is the glass half empty, or half full?" Then I think about how far we have come to better understand the genetic disorder fragile X. It seems that it is not such a frightening part of our lives. We are so

blessed to live in this age of modern technology and medical professionals willing to work in research so that we can receive the most recent developments in treatment, educational tools, and gene research. We are also blessed with the dedicated administrators, teachers and therapists that provide the physical and educational assistance to our children, grandchildren and great-grandchildren.

### **The Future**

What lies ahead we do not know, but with God's help and family support, we'll accept and face the challenges as they come. We are determined to pass on the importance of learning about fragile X syndrome. We continue to urge extended family members to be tested. We will pass on the information we have accumulated and steer them to websites and books now available. Most of all we want to share with others our optimism concerning the research that is in progress everyday. We intend to press family members to make a donation, small or large, to the organizations associated with fragile X so they can continue to study on our behalf.

### *Summer Research Fellowships*

*Funded By The NFXF's Research Funds*

## **CALL FOR PROPOSALS**

The National Fragile X Foundation will fund 4-6 summer research fellowships at \$2,500 each through the NFXF Research Funds. Proposals must be submitted to the NFXF by April 22, 2002. Notification of award will be made in May 2002. The proposals should be 3-5 pages, describe the research project in depth, and the role of the summer fellow and the mentor. The CV of the mentor and the fellow should be included as an addendum. The fellow can be an undergraduate or graduate student and the mentor should be experienced in fragile X research.

The research proposal should advance our knowledge in the field of fragile X including molecular genetics, genetic counseling, cytogenetics, screening, psychology, pharmacotherapy, educational intervention, computer technology, vocational intervention, or other areas that advance our knowledge regarding the biology of individuals with fragile X syndrome.

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## A Practical Resource for Teachers. We need your help!

The National Fragile X Foundation's new two-year **Education Project** will result in the creation of a resource binder filled with practical ideas and strategies for teachers and parents. The focus of the project is the education and inclusion of children with fragile X in preschool through high school classrooms. Over the next two years we will also develop web-based resources along with the published binder of information. The Education Project needs your help!

We are asking parents, teachers, therapists and others to send in their successful adaptations of lesson plans, curriculum, classroom setup or teaching approaches. What are some of the techniques that have been used with success, to teach reading, writing, mathematics, conversation skills, socialization skills, social studies, vocational skills, science, or other subjects? What materials have been found to be successful in teaching students with fragile X, whether published educational programs, games, software, or other materials?

We are also requesting nominations for "Master Teachers" who will present at a panel during the National Fragile X Foundation's 8th International Conference in July, 2002. Master Teachers are those teachers who have experience successfully adapting their teaching lessons to accommodate children with fragile X. At the Chicago conference we will bring together successful teachers from all over the country who will share their ideas and insights. We plan for many of their lesson plans and adaptations to be included in the resource binder and on the website.

Please submit the form below, or feel free to contact us with your ideas, suggestions, success stories, or materials. We particularly look forward to hearing from parents regarding materials and strategies that have worked for your children!

**CONTACT:** *Carolyn Ybarra, Ph.D.*  
*Education Project Coordinator, NFXF*  
*carolyn.ybarra@stanford.edu*  
*(Please put "Education Project" on subject line)*

### Fragile X Education Project Submission Form

Name: \_\_\_\_\_

Address: \_\_\_\_\_

City/State/Zip: \_\_\_\_\_

Phone/Fax: \_\_\_\_\_

Email: \_\_\_\_\_

I nominate the following teacher as a "Master Teacher" of children with fragile X. This teacher has successfully created or adapted materials for their students with fragile X. (You may self-nominate).

Master Teacher Name: \_\_\_\_\_

Address: \_\_\_\_\_

City/State/Zip: \_\_\_\_\_

Phone/Fax: \_\_\_\_\_

Email: \_\_\_\_\_

Reason for nomination (what has this teacher done in their classroom for children with Fragile X?): Please attach second sheet if necessary: \_\_\_\_\_

I suggest the following resources, materials, lesson plans, or software for use with children with Fragile X (Please attach copies as appropriate): \_\_\_\_\_

Name of material: \_\_\_\_\_

Subject area (math, reading, writing, etc.): \_\_\_\_\_

Approximate grade level for this material, if applicable: \_\_\_\_\_

Ordering information or author and contact information: \_\_\_\_\_

Description of how this has been used in your experience: \_\_\_\_\_

*Feel free to submit or discuss ideas that might not fit into this form.*

Please submit to project coordinator Carolyn Ybarra at:

**National Fragile X Foundation**  
**Education Project**  
**P.O. Box 190488**  
**San Francisco, CA 94119**

Email: [carolyn.ybarra@stanford.edu](mailto:carolyn.ybarra@stanford.edu) (Please put "Education Project" on subject line)

Fax: 650-367-7571

## It's Not If, It's How and When

### *Toilet Training In Less Than A Century*

By Jeffrey Cohen

**Potty training.** Two words that strike fear in the hearts and minds of all parents of children with fragile X. The neighbor's kid did it at two and they can't stop talking about it. Your older child did it at three and your nephew from Cleveland did it at four, all by himself without any prompting. But your son or daughter is five, no six, seven, or maybe even ten or twelve and still shows no interest. This is beginning to get embarrassing and depressing. What's a parent to do?

Let me first tell you that I'm not a doctor, a psychologist or a behavioral specialist. I'm *just* a parent of children with fragile X. My thoughts are based on my own personal experience, consultation with many "experts," reading, but most importantly on common sense, creativity, "thinking outside the box," and a passionate desire to relate to, interact with, and help my child. In other words, tools that we all have.

First, avoid the mistake that so many others make when interacting with our children. Just like typical kids are all different, so too are all kids with fragile X. They don't all learn the same way or at the same pace and often different and creative approaches are required to learn new skills. The point is that whether you use the approach detailed here or elsewhere in many other good resources *don't feel like you have to follow the instructions to the letter*. Nobody knows your child better than you. Nobody knows how they learn, what can grab and hold their attention, when something is working and when something isn't. After reading this proposed method and any others you find, sit down and really think about how you can modify and adapt the procedure to fit your child.

In our case we started with resources that are often recommended: "*Toilet Training in Less Than a Day*" by Nathan Azrin and Richard Foxx, and "*Once Upon a Potty*" video—both checked out from our public library. The book recommends using a doll as a role model and we decided right away that would be far to abstract for

our son (4.5yrs) so we decided that I would be the role model.

Before reviewing the procedure, first a word on when to begin this process. Remembering again that I do not have a medical or psychological background, I believe that when you are able to interact successfully and have fun with your child, on their level, that it's a good time to start. What I'm talking about is based upon what I still believe was some of the best advice I ever received from any professional about my son. He was evaluated by a psychologist at about age 4 before we even had the diagnosis of fragile X. At an early session he put my son and I in an empty waiting room with a hidden camera to watch us interact. Some toys, chairs, magazines and a tall corn plant, that was it. I tried to get my son to play appropriately with the toys but he had his mind set on that corn plant. Batting at the leaves was to be the activi-

ty of the day and I had to decide whether to fight it or play along. I decided to play along and thus was born the "corn plant game". He'd bat a leaf, then I. He'd bat two, then I'd bat two. Pretty soon we were taking turns, setting the rules, laughing, interacting and having a great time. The psychologist loved every minute of it (the plant could be replaced) and explained that my son and I had stumbled onto what he thought was the secret of reaching him regardless of the diagnosis. Don't try to

forcefully pull him out of his world, go in there with him and begin the interaction on his level with an activity of his choosing. Begin a pattern of interaction and turn taking and eventually the transition to more appropriate activities and interactions will follow.

As it turns out this guy was right on the mark. This was the first of many great games to follow. The "lumpy couch" game and the "jumbling three of anything" game are two that come to mind. The activity was chosen by my son and then we made a fun, laughing, turn taking game out of it.

Which brings us back to potty training. The basic method was to take the procedure from start to finish and break it down into small component parts. Getting up off the couch, walking to the bathroom, opening the door, turning on the light, pants down, sit and go. Each

**The procedure calls for all the salty snacks they can eat followed by all the liquid then can drink. The idea is to keep the bladder full at all times to maximize opportunities.**

*Continued, page 20*



# 8th International Fragile X Conference, July 17-21, 2002

*Sheraton Chicago Hotel & Towers, Chicago, Illinois USA*

## CONFERENCE REGISTRATION FEES

Note: Fees for all registrants include the luncheon and the banquet dinner.

### REGISTRATION FEES FOR **CURRENT NFXF BASIC OR ASSOCIATE MEMBERS**

Type	Early Registration by 5/17/02	Regular Registration 5/18/02-6/30/02
Professionals	\$390	\$415
Family (2 Parents)	\$380	\$405
1 Parent	\$250	\$275
Student	\$250	\$275

### REGISTRATION FEES FOR **CURRENT NFXF PROFESSIONAL, FAMILY, SUPPORTING, DIAMOND, and LIFETIME MEMBERS\***

Type	Early Registration by 5/17/02	Regular Registration 5/18/02-6/30/02
Professionals	\$370	\$394
Family (2 Parents)	\$361	\$385
1 Parent	\$237	\$261
Student	\$237	\$261

\* The above fees include the 5% discount for current NFXF members. (Not including "Basic" or "Associate")

### REGISTRATION FEES FOR **NON-MEMBERS\***

Type	Early Registration by 5/17/02	Regular Registration 5/18/02-6/30/02
Professionals	\$415	\$440
Family (2 Parents)	\$405	\$430
1 Parent	\$275	\$300
Student	\$275	\$300

\* The above fees include a \$25.00 non-member fee

An additional \$25.00 late-fee will be assessed for all registrations made or postmarked after June 30.

### **SINGLE-DAY REGISTRATION**

Single-day registration is \$100 for all NFXF member types and \$125.00 for all non-members. Single Day registration does not include the Thursday luncheon or the Saturday dinner banquet. A limited number of meal tickets may be available for purchase at the NFXF table on the day of those events.) Note: Wed. & Sun. are half days and 50% off.

### **NOT A MEMBER?**

#### **NOW IS A PERFECT TIME TO BECOME ONE!**

MEMBERSHIP conference discounts (5%) will be honored for all memberships made at the time of registration. Become a member of the National Fragile X Foundation and receive the Foundation Quarterly while saving money at the conference and on resource materials. (See the NFXF Membership form for details on additional membership benefits.)

#### **Membership Types**

Basic: \$25.00	Supporting \$100.00
Professional \$40.00	Diamond \$500.00
Family \$60.00	Lifetime \$5000.00

## We're Proud To Be The Official Airline For The National Fragile X Foundation

Meeting attendees can enjoy 5% off the lowest published fare or 10% off the full coach fare on American. Plus, receive an additional 5% discount when you purchase your tickets 60 days in advance.



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The National Fragile X Foundation.

Call American At 1-800-433-1790

Or ask your Travel Agent

And Ask For STARfile 0462BB.

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# 8th International Fragile X Conference, July 17-21, 2002

Sheraton Chicago Hotel & Towers, Chicago, Illinois USA

## CONFERENCE REGISTRATION FORM

Please print clearly!

### REGISTRATION INFORMATION

Name(s): \_\_\_\_\_  
Organization (if applicable): \_\_\_\_\_  
Title: \_\_\_\_\_  
Address: \_\_\_\_\_  
City: \_\_\_\_\_ State: \_\_\_\_\_ Zip/Postal Code: \_\_\_\_\_  
Country: \_\_\_\_\_  
Phone: \_\_\_\_\_  
Fax: \_\_\_\_\_  
E-mail: \_\_\_\_\_

### REGISTRATION MEMBERSHIP STATUS

Please refer to the Conference Registration Information opposite, as you complete the following. Be sure to check the appropriate box.

- I/We am/are registering as a **current** NFXF **Basic** or **Associate** member
- I/We am/are registering as a **current** NFXF **Professional, Family, Supporting, Diamond** or **Lifetime** member
- I/We am/are registering as a **non-member**
- I/We am/are registering as a **current** member for **single-day registration** for the following days (circle): **W / Th / F / S / Su**
- I/We am/are registering as a **non-member** for **single-day registration** for the following days (circle): **W / Th / F / S / Su**

### CHILDCARE

**Free** for those registered at the Sheraton Hotel— All others at the childcare door: \$20 per half day, \$30 per full day

I/We anticipate needing childcare Number of children \_\_\_\_\_ Ages \_\_\_\_\_

(Pay at door)

### MEALS

Fees for all registrants include the luncheon and the banquet dinner. Separate meal tickets available for children and guests.

I/We am/are purchasing \_\_\_\_\_ additional meal tickets for the **Thursday, 7/18 luncheon at \$45 per adult, \$25 per child.** \_\_\_\_\_

### BANQUET

Saturday evening, July 20: Dinner/Banquet, Silent & Live Auction and Entertainment by American English, the premier Beatles impersonator band in the world.

*Sam Leach, original promoter of the Beatles and founder of the Merseybeat movement says: "American English is the best Beatle band on the planet. Lightning has struck twice, first with the Beatles and now again with American English!"*

Fees for all registrants include the banquet dinner and luncheon. Separate meal tickets available for children and guests.

I/We am/are purchasing \_\_\_\_\_ additional meal tickets for the **Saturday, 7/20 banquet at \$85 per adult, \$25 per child.** \_\_\_\_\_

### DINNER CRUISE

Friday, July 19: Join families and faculty while experiencing the world renowned Chicago architecture and skyline from Lake Michigan aboard our exciting and entertaining dinner cruise. This event should not be missed! (7-10:00 PM.) Includes dinner and dancing!

I/We am/are purchasing \_\_\_\_\_ tickets for the **Friday, 7/19 7:00-10:00 PM Dinner Cruise at \$65 per person.** \_\_\_\_\_

**PAYMENT** Please include meal ticket and dinner cruise fees. (Childcare fees are payable at the door.)

Based on the registration fees, calculated from Fee Schedule, and any other fees:

**Total amount due \$** \_\_\_\_\_

Payment in full must be received by June 28, 2002. Cancellations through June 28, 2002 will be refunded in full less a \$25.00 administrative fee. No refunds will be available after that date.

Check enclosed:  check number \_\_\_\_\_ check amount \_\_\_\_\_

Credit card payment:  VISA or  MasterCard Card number \_\_\_\_\_

Exp. date \_\_\_\_\_ Total to be charged \$ \_\_\_\_\_ Cardholder's signature \_\_\_\_\_

*By signing here, you authorize the NFXF to charge the above amount to your card.*

Please contact the National Fragile X Foundation directly with any special needs or requests at 1-800-688-8765.

**Mail to: National Fragile X Foundation, PO Box 190488, San Francisco, CA 94119-0488 or FAX to: 925-938-9315**

## It's Not If, It's How and When

*Continued from page 16*

activity is done by the role model and then the child. The book said use a doll for a role model but in our case I was the doll. I exaggerated every step and turned each into the kind of game my son and I frequently played. We took turns getting up off the couch and doing each successive step and when my son took his turn and completed each step he was greeted with absolutely wild praise and glee. He loved it. For actual successes we had a celebration fit for a king's coronation.

The procedure also calls for all the salty snacks they can eat followed by all the liquid they can drink. The idea is to keep the bladder full at all times to maximize opportunities. After a success, when you're back on the couch watching the video, check the pants. (Dry pants inspection game.) If they're dry it's time for more wild celebration.

Have plenty of underpants on hand for accidents. Each time one happened he had to change himself and put his dirty laundry in the washing machine. Accidents also set up another game: the walking through the route to the nearest bathroom from the location of the accident game.

In between all this merriment keep the pretzels and pop flowing and watch the video about potty training. No other videos or TV. No other topic for the day or in our case for the entire weekend. It might even be a good idea to let your other kids sleep out. From the time we woke up on Saturday to the time we went to bed on Sunday this is all we did—and it worked. After 2 days we were 75-80% complete. The procedure finished gradually over the next 6 months but we never went back to diapers. Keep a close eye and continue to heap wild praise for success with dry pants inspections.

You might feel a bit silly about all of this and if you're embarrassed pull the shades. But to succeed you'll need to tap into your inner child and have a good time with it. My wife and I still laugh out loud every time we re-tell the story.

Now you know what they say about "always" and "never" so results in every case cannot be guaranteed, but I strongly believe that with the right mix of perseverance, creativity and unbridled fun it's not a question of if, only how, and when. Good Luck!

**Editor's Note:** Jeffrey Cohen is President of the National Fragile X Foundation and the father of two children with fragile X. His description of his successful experience with his son has appeared on the internet and has been shared amongst many parents. It is meant to be just one parent's description, and is printed in this publication as a reminder to parents that a Ph.D. in special education is not required to toilet train your child with special needs. Nonetheless, many of the specialists do have a great deal to offer in the way of experience, suggestions and insight into the psychological and physical needs of your child.

In addition to the resources mentioned by Jeffrey, we recommend, *Fragile, Handle With Care*, by Marcia Braden, Ph.D., and *Fragile X Syndrome, A Parents Guide* by Jayne Dixon-Weber. Both titles provide very good overviews of the issues regarding toilet training.

As additional sources of information, we suggest an internet search using "toilet training special" as your search text. There you will find many interesting and helpful discussions, suggestions and lists of resources including some specialized products.

As always, a word of caution when searching the internet: Always look for information provided by trained and/or experienced individuals. Be skeptical of claims that seem to defy common sense. Talk with parents of children who have special needs similar to your child's. Most importantly, always check with your doctor before embarking on a new potty training procedure. You always want to rule-out the possibility that physical problems are interfering with your child's ability to develop bladder and bowel control.

### Mission Statement of the National Fragile X Foundation

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 syndrome.

# CONTINUE YOUR SUPPORT OF FRAGILE X

## RENEW YOUR ANNUAL MEMBERSHIP WITH THE NATIONAL FRAGILE X FOUNDATION!

Your annual membership with the National Fragile X Foundation helps spread the word about fragile X syndrome while also helping the Foundation to provide free information and materials, referrals to support groups and support for research. All members receive 4 copies annually of our informative newsletter, *The Foundation Quarterly* in addition to these membership benefits:

### Lifetime Membership - \$5,000

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\* The fX Files: A comprehensive, continually updated compilation of over 150 pages of articles on a variety of topics related to fragile X research, medical interventions, education, and more. Available in print or as a PDF file (floppy disk or email attachment)

\*\*The FragileX.org Website CD: A portable website—our entire website with over 200 pages of valuable information including back issues of the newsletter and a 4 minute informational video

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# CALL FOR ABSTRACTS

## 8th International Fragile X Conference

Chicago, Illinois, July 17-21, 2002

This is a call for abstracts to be submitted for the 8th International Fragile X Conference. Submitted abstracts must be completed and sent to the National Fragile X Foundation no later than May 17th, 2002. All abstracts must be submitted electronically. Please visit the "Foundation" page at [www.FragileX.org](http://www.FragileX.org) where there will be a link to the Abstract Submission Form. Be certain to indicate the number(s) of the workshop(s), listed below, for which you are submitting an abstract for presentation. The Foundation will respond directly to the first named presenter, listed on your abstract, to advise of acceptance or refusal. The two presentation formats are slide/PowerPoint, or poster. Slide/PowerPoint presentations occur within individual sessions and last between 5-10 minutes each. All poster presentations take place at a single, designated time, over a 1- to 2-hour period. Please note: If you are presenting an abstract, you must register for the conference.

The purposes of the 8th International Fragile X Conference are: 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 and related forms of X-linked mental retardation.

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 will include 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, and 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 & Clinical Advisory Committee, and membership. Conference format will include symposia, lectures, workshops, panel discussions, and poster presentations. All events are scheduled at the Sheraton Chicago Hotel & Towers. Conference programming will run from 8:00AM to 5:00PM daily. Additional social and leisure activities will be available in the evenings.

### WORKSHOP TOPICS

- |   |  |   |
|---|--|---|
| 1. FMRP & Molecular Clinical Correlations | 13. Issues for Premutation Carriers            | 25. Augmentative/Alternative Communication    |
| 2. Gene Therapy                           | 14. Neurological Problems in Pre/Full Mutation | 26. Occupational Therapy/S.I.                 |
| 3. Molecular Studies in FX                | 15. Neuropsychological Studies                 | 27. Multidisciplinary Interventions           |
| 4. Mouse Models                           | 16. Psychopharmacology                         | 28. Academic Interventions                    |
| 5. Synaptic Structure & Function          | 17. Screening & Prevalence Studies             | 29. Inclusion/Mainstreaming Issues            |
| 6. X-Linked Mental Retardation            | 18. Genetic Counseling Issues                  | 30. Spiritual/Faith-Based/Cultural Issues     |
| 7. Medical & Neurological Issues          | 19. Adult Issues                               | 31. Financial/Insurance/Planning Issues       |
| 8. Electrophysiological Studies           | 20. Emotional & Behavioral Issues              | 32. Mother/Fathers/Sibling/Grandparent Issues |
| 9. Growth and Endocrine Issues            | 21. Interventions for Behavior/Aggression      | 33. Other:                                    |
| 10. Reproductive Options for Carriers     | 22. Sexuality Issues                           |   |
| 11. Neuroimaging                          | 23. Speech & Language Studies                  |   |
| 12. Association Between Autism and FX     | 24. Speech & Language Interventions            |   |

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