



The National **Fragile X** Foundation

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Don't forget! The National Fragile X Foundation's 8th International Fragile X Conference, Chicago July 17-21, 2002

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.

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 syndrome.

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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

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Genes, Schools and Family:

What Influences Cognitive and Behavioral Outcome in Children with Fragile X Syndrome?

David Hessel, Ph.D., Jennifer Dyer-Friedman, Ph.D., Bronwyn Glaser, B.A., Heather Erba, Ph.D., and Allan L. Reiss, M.D. from the Behavioral Neurogenetics Research Center, Department of Psychiatry and Behavioral Sciences, Stanford University

The research described below was presented at the International Fragile X Conference in Los Angeles in July 2000 and discussed in the Spring 2001 edition of this newsletter. At that time, the results were preliminary because data collection was incomplete. The current article is a summary of three of several published articles thus far based on the completed project.

Since the discovery of the fragile X mental retardation (*FMR1*) gene a little over a decade ago, there has been much scientific progress in understanding the biological and genetic roots of fragile X syndrome. In a relatively short period of time, researchers have made important breakthroughs in understanding the molecular genetics and neurobiology of fragile X. Like a painter's canvas, the complete picture of this subject of intense study has become clearer over time as material is added to different areas and we step back to see the entire field of view. The scope and clarity of this biological picture is essential for developing effective treatments for children with fragile X in the future. Yet we know that each child's experience is determined by more than his or her *FMR1* gene. Most of us believe that positive family experiences, effective school programs, and a supportive community contribute to each child's constructive development. However, because the cognitive ability, emotional adjustment, and practical day-to-day skills are significantly affected by the fragile X gene, we may focus on the biological basis of the disorder and either forget or make assumptions about environmental influences on development.

At the Behavioral Neurogenetics Research Center (BNRC) at Stanford University, directed by Dr. Allan Reiss, we posed the following research question: "What influences the cognitive, behavioral, and practical day-to-day skills of children with fragile X syndrome?" More specifically, "Why are some children with fragile X more affected than others?" One possible explanation for the differences among children is the protein produced by the *FMR1* gene (FMRP). We know that this protein is critical for normal brain development and function. For example, it may help with the process of nerve cell development and function and play an important role in the efficiency of communication between nerve cells. But due to abnormalities in the function of the gene, FMRP is diminished to varying degrees in children with fragile X. So, a plausible answer to the research question is that children whose cells express more FMRP have stronger cognitive skills, fewer emotional or behavior problems, and in general have better outcomes. But, beyond this, once we account for the likely impact of the genetic abnormality, does the child's environment, for example the characteristics of his home, his/her parents, school or therapeutic program, also contribute to how he/she is learning and developing? Is it possible that these environmental characteristics help compensate for the deficit in FMRP? We know from previous research that these environmental factors influence typically developing children. In regard to children with fragile X, the answers to these questions are especially important

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Genes, School and Family...

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because they will help us develop treatment programs for the present, that will focus on aspects that are most likely to lead to positive changes.

To examine these questions, the staff of the BNRC visited the homes of 120 families in 36 states and Canada. Each family had at least one child with fragile X and, for comparison purposes, one unaffected sibling. The full day visit included cognitive testing of children and parents, behavioral evaluation of children through direct observation and parent reports, information about parental psychological health and stress, measurement of the effectiveness of educational and therapeutic services, and a standardized assessment of the home environment. (A hormone associated with stress also was examined in conjunction with a socially challenging task. See the reference below for details on this part of the project). First, three central child characteristics of development were chosen for examination: cognitive ability (IQ), behavioral/emotional problems, and adaptive behavior (i.e., “practical” communication, socialization, and daily living skills necessary for independent functioning and coping). Next, based on the scientific literature pertaining to fragile X, typical development, and our specific hypotheses, potential contributors to the children’s development were identified. These possible contributors included: *FMR1* protein expression, the child’s age, the effectiveness of educational and therapeutic services provided to the child, parental cognitive skills and psychological health, the overall quality of the home environment (for example safety, learning enrichment, parent responsiveness, organization), and family economic status (a measure of financial resources). Finally, statistical tests were used to identify the biological or environmental variables that contributed most significantly to the children’s cognitive abilities, behavior, and life skills.

Cognitive Ability: What contributes most to the cognitive ability of children with fragile X? First, as might be expected, the heritability of IQ played an important role. That is, some parental cognitive strengths and weaknesses are passed on to a child whether or not he has fragile X. The heritability of intelligence appeared to be stronger for girls than for boys with fragile X. Beyond that, as expected, the fragile X protein (FMRP) played a significant role for both boys and girls with fragile X such that increased

expression of FMRP led to higher IQ scores. Cognitive skills pertaining to attention and math computation were most affected by FMRP levels. Finally, among environmental characteristics, only quality of the home environment contributed to cognitive ability. Children living in safer, more organized, enriched, and responsive home environments had higher IQ scores. (Interestingly, this was specific to verbal cognitive and attention skills only, not visual-perceptual ability.) The impact of the home environment was independent of economic status, as family income was not associated with any measure of child cognitive ability.

Behavior Problems: Which characteristics best predict the severity of behavior problems in children with fragile X? Here, a somewhat different story emerged. First, FMRP was found to play a role, but its influence was limited primarily to what are referred to as “internalizing problems” (withdrawn, anxious, or depressed behavior), in contrast to “externalizing” problems (aggression, oppositional behavior), predominantly in girls. Second, the impact of the home environment was limited to autistic symptoms such as gaze aversion, social problems, and repetitive behaviors in boys with fragile X. In other words, boys living in more effective home environments had fewer autistic symptoms. Finally, boys with fragile X receiving more effective educational and therapeutic services had fewer of all types of behavior problems.

Communication, Socialization, and Daily Living Skills: Which characteristics best predict the practical day-to-day skills children develop? In our results, the child’s cognitive ability was particularly important as a determinant of development. As one might expect, children with fewer cognitive resources had more difficulty coping with life challenges such as daily living skills and social interactions. Thus, in this analysis, child IQ was a strong and consistent determinant of practical skills in both boys and girls with fragile X. In fact, IQ accounted for 38% of the individual differences in practical skills in boys and up to 64% in girls with fragile X. In the group of boys, we again saw the impact of the home environment, which accounted for another 20% of the individual differences in practical skills. Effectiveness of services, family income, and parental psychological health had no impact on practical skills development of children with fragile X.

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Finally, Everyone Knows About Fragile X!

Got your attention, didn't I? Wouldn't it be great? No more, "fragile what?" Our mission of spreading **awareness** finally achieved? True, our public service announcement for television has now been seen by over 10 million viewers. True, CBS has approved it for a national airing. And it's true that we have more resource groups, more parent contacts, and are answering more phone calls and emails than ever before. It's even true, at least based upon our interactions with physicians around the country, that more doctors know about fragile X and are requesting the test.

But then the phone rings. It's the mother in a suburb of a mid-sized Texas city who, until she saw our PSA on television, really thought that she and her 13 year old son were the only ones affected by this "rare" condition. Or it's the doctor in a major urban hospital whose own teenage daughters have just been diagnosed with fragile X, a condition even he had never heard of. Maybe it was the email from the lonely mother in Poland who feels like she's up against an entire medical establishment that seems to have never heard of the syndrome. Or maybe it's just the nagging notion that even if we take all the names in our database, add all the people who have attended our conferences and add all the contacts who email or call, that there may still be thousands of families laboring under the impression that they are the only ones and with nowhere to turn for help.

This is why your help with promoting our PSA on your local television stations, your help in organizing

awareness and fundraising events, your help in talking to your friends, neighbors, teachers, doctors, etc., etc., etc., makes a world of difference. So keep it up! You are making the world a better place for all whose lives are touched by fragile X. (And while you're at it, take a look at our list of parent contacts and resource groups and centers. If there is one in your area, join. If nothing exists, start one. We'll help!)

What we hope will be another key component of our education and awareness efforts is the National Fragile X Foundation's **Education Project** which is well underway. (see page 17) I cannot emphasize enough how important this lesson planning guide will be to families and teachers, and as it develops we are determined to keep it practical and easy to use. We realize that teachers are very busy people and as much as we sometimes want their students with fragile X to be their only concern, we realize that is not the reality. Therefore, an easy-to-use resource, that can be used to quickly make modifications in a student's daily educational experience, is one of our primary goals.

On the **research** front, please take the time to read the reports from the recipients of our fellowship awards in 2001. These individuals represent part of the future of fragile X research and the National Fragile X Foundation is pleased to be a part of that future. The Foundation supports research in many ways including:

The William Rosen Research Fund which was established in 1995 by the National Fragile X Foundation

and the Rosen family. (Bill's daughter and son in law, Arlene and Jeffrey Cohen are members of our Board of Directors) William Rosen was diagnosed with mosaic fragile X at age 75. He lived a wonderful and successful life as a husband, father, grandfather, businessman and decorated hero during World War II. Following his death, his vital organ tissue was donated by his family to be used for research into fragile X.

The Harry LeCover Memorial Research Fund was established in 1999 to honor the life of Harry. Harry LeCover was the grandfather of Daniel LeCover who has Fragile X. Upon Harry LeCover's passing at the age of 85, his son Stephen and daughter-in-law Deborah requested that any donations made by family and friends be donated to The National Fragile X Foundation in his memory.

Additional funds come from the Foundation's **Special Research Fund** which exists to fulfill that part of our mission to: "advance research toward improved treatment and a cure for fragile X syndrome." The NFXF also supports and promotes research through our **International Conference Series**. Through the Foundation's considerable organizational and financial efforts, scientists and researchers from around the globe gather to lecture, share, and meet, to strategize their next steps. From these face-to-face gatherings flows the excitement and challenge which is the basis of all good scientific endeavors. When doctors, therapists, parents and teachers are added to the mix the result is truly unique.

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Xtraordinary

Accomplishments

My Son Spencer

Mary Lee Shelton

My son Spencer Shelton is an 11-year-old 5th-grader with Fragile X Syndrome whose greatest love is basketball. His team, the All Stars, practices every day after school and plays games most Saturdays. Like other children with fragile X, Spencer struggles with social skills, so staying active and busy with extra-curricular activities provides him with much-needed social interaction and a built-in social calendar. In addition to basketball Spencer dances with SACBE, a Ballet Folklorico troupe that practices three nights each week. He attends Sunday School and performs with a handbell choir at church. He has been in Cub Scouts all through elementary school, and presently contemplates joining Boy Scouts in middle school.

These activities bring spin-off events. Spencer is now rehearsing the part of Simba for a performance of *I Just Want To Be King* in a church musical. He will perform for his school talent show a Ballet Folklorico dance to the music from *Spy Kids*.

At school Spencer studies reading, math and social studies in a self-contained classroom. He attends science, music, art, library, physical education, lunch and all field trips with the regular education students. Spencer tests at the high second-grade reading level, but functions in the classroom at the third-grade reading level. He has mastered addition and subtraction using Touch Math. Multiplication tables have been memorized but he has not yet tackled division. He just aced an oral social studies test about the presidency during which he explained, among other things, the characteristics and meaning of the presidential seal and the definition of the constitution.

These are some of the positive things about Spencer, but like other individuals with fragile X, he struggles in many areas. Although he practices basketball relentlessly, he is the weakest player on his team. This bothers him immensely, so much so that as soon as he got home from school recently, he demanded to write President Bush. He told Bush that he (Spencer) “stinks at basketball,” and that he would like to meet the president so Bush could learn more about fragile X, give more money to research, and find a cure so that Spencer could be better at basketball. He has yet to receive an answer.



Spencer Shelton, top left, with his All Stars teammates. For Spencer teamwork has been the key to his success.

Spencer has trouble hearing the beat and trouble using the correct foot or arm in dance class. When he began dance, the teacher (who does not speak English) was very frustrated with Spencer. Through an interpreter I explained that he had fragile X. Because Spencer doesn't look or act differently (generally speaking) than other children, people tend to think I'm a defensive mother, making “excuses” for him. So I e-mailed Robby Miller and asked for a Spanish language book to explain fragile X. He promptly sent the book, and what a difference it made! I saw more patience and compassion for Spencer. In about a month the teacher told me (again through an interpreter) that

he admired my husband and me very much for what Spencer had accomplished. He said he knew it took the efforts of many people working together to get to this point, and that he wanted to be a part of that team.

His dad and I are the captains of his team. His 16-year-old brother and 14-year-old sister (both unaffected) are his primary role models. His gifted and dedicated teacher (Sonya Vestal), his principal, music teachers, basketball coach, church leaders and extended family members and friends and physicians like Randi Hagerman and Robert Woody are all key team players. For Spencer teamwork has been the key to his success.

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EDITOR'S NOTE: The above article is the first in a new section of the Foundation Quarterly designed to highlight the accomplishments of individuals with fragile X. To have your child's accomplishments considered for publication, please submit an article of 300-800 words along with a photo suitable for publication. Accomplishments in any area (sports, recreation, education, employment, family, civic, etc.) are encouraged. Contributions can be submitted via email (preferred) or by postal mail to:

*Xtraordinary Accomplishments
National Fragile X Foundation
PO Box 190488
San Francisco, CA 94119.*

Please note: The NFXF reserves the right to edit articles, as appropriate, for clarity and space purposes.

Announcing the 4th in a Series of Free, NFXF Sponsored

Question & Answer Online-Audio Webcasts with Fragile X Specialists

The National Fragile X Foundation presents
Elizabeth Berry-Kravis, MD, Ph.D., Online
Tuesday, June 4, 2002

5:00PM Pacific 6:00PM Mountain
7:00PM Central 8:00PM Eastern

Elizabeth Berry-Kravis MD, PhD., is an Associate Professor of Pediatrics, Neurology, and Biochemistry at RUSH-Presbyterian-St. Luke's Medical Center in Chicago. She received her Bachelor of Science degree from the University of Notre Dame, her Ph.D. in Biochemistry, M.D. from the University of Chicago, and completed her Pediatrics residency and Child Neurology fellowship also at the University of Chicago. She established a Chicago-area Fragile X Clinic in 1992 and currently sees about 200 patients with fragile X syndrome for management of neurological, medical, and behavioral issues. In addition, she co-directs the Molecular Diagnostics Laboratory at RUSH including fragile X DNA analysis. Dr. Berry-Kravis will be one of the featured speakers at the 8th International Fragile X Conference in Chicago this summer.



If you would like to ask Dr. Berry-Kravis a question, please call 1-800-218-0204 at the time of the event, or, email questions to NATLFX@FragileX.org with a subject heading of "for Dr. Berry-Kravis."

**To participate in the online session, visit
www.FragileX.org**

Click on the "On Line Q&A" icon for free registration. An internet connection and computer speakers are required to participate. Participants are encouraged to register and test their connection in advance of the event. Additional instructions will be provided once you have registered.

Is it Autism or Fragile X?

Randi Hagerman, MD

EDITOR'S NOTE: Many parents contact the National Fragile X Foundation confused about their child's diagnosis. On the one hand, they've been told that their child has autism, "autistic spectrum disorder," or some degree of autistic-like characteristics. In addition, they've now been told that their child has fragile X syndrome or that he or she is going to be tested for fragile X. This can add an unnecessary burden to those already struggling to understand their child's special needs. (see Brenda Finucane's article,

"Diagnostic Alphabet Soup—Demystifying Genetic and Behavioral Labels" in the Winter 2001 issue of the Foundation Quarterly.) As a result, we have asked Dr. Hagerman to

write this article. Dr. Hagerman is chair of the NFXF's Scientific & Clinical Advisory Committee, and the Medical Director of the UC Davis Medical Center's M.I.N.D. Institute, which focuses on neurodevelopmental disorders in children and adults including autism and fragile X.

Most children with fragile X...are interested in social interactions and do not meet the... criteria for autism. However, a subgroup...does meet the criteria for autism

The association between autism and fragile X was first reported by Brown et al. (1982) and was subsequently confirmed by many others leading to an extensive field of research. In discussing this association it is important to remember that autism is defined behaviorally using the criteria of the DSM IV manual which include lack of social reciprocity or responsiveness, abnormal use of language and communication, and a restricted repertoire of activities and interests. Autism is a heterogenous disorder which means that there are several known causes of autism including phenylketonuria (PKU), tuberous sclerosis and 15q duplications. However fragile X is the most common known cause of autism so far identified. Autism is strongly genetic and it is likely that the inheritance of multiple genes predisposing an individual to autism is necessary in many cases for the full behavioral syndrome to be manifested.

The typical features of fragile X syndrome (FXS) i.e. hand biting, hand flapping, poor eye contact, shyness, and social anxiety are probably related to the sensory hyperarousal that has been documented by many investigators including Belser and Sudhalter (1995), Miller et al. (1999), and Roberts et al.(2002). These features are often also referred to as *autistic-like* features because they can be seen in individuals who have autism without fragile X. *Most children with fragile X, however, are interested in social interactions and do not meet the diagnostic criteria for autism.*

However, a subgroup of children

with fragile X do meet diagnostic criteria for autism. Over the last decade many studies have evaluated this issue and the percentage of children with FXS who have autism has varied from 15 to 33%, mainly because the diagnostic criteria for autism has varied and the diagnostic tools used have changed.

Recent work by Don Bailey and colleagues has found that in young boys with FXS, 25% met the criteria for autism using the Childhood Autism Rating Scale (CARS) and that their profile of behaviors was very similar to that of children with autism and without fragile X. They also found that children with autism and FXS together, had a lower IQ than children with either FXS alone or autism alone (Bailey, Hatton et al. 2001). Furthermore, the level of the fragile X protein (FMRP) did not correlate with the presence or absence of autism. (Bailey, Hatton et al. 2000) This suggests that autism with fragile X may relate to additional genetic or environmental factors that could be additive to the FMR1 mutation.

Our studies also agree with this hypothesis. We recently reported a comparison study of preschoolers with FXS, and age matched children (controls) with autism but without fragile X and another set of children (controls) who had developmental disabilities but without autism or FXS. (Rogers, Wehner et al. 2001) We evaluated all of these children with what are the agreed gold standard diagnostic tools for autism including the Autism Diagnostic Observation Schedule-Generic (ADOS-G) and the Autism Diagnostic Interview-Revised (ADI-R) and also utilized the the DSM

IV criteria, IQ and adaptive behavior measures.

We found that 33% of the children with FXS met the full criteria for autism. Their profile of autistic features was indistinguishable from the children with autism without FXS.

A Parent's Perspective on Autism and Fragile X

Our seven year old son has a dual diagnosis of fragile X syndrome and autism. For our family, the autism diagnosis has created additional opportunities for understanding and treatment. First of all, most educators are at least familiar with autism (unlike fragile X syndrome, but we're working on that). This familiarity helps schools more readily understand the challenges and needs our kids typically face and how they can be accommodated. Also, because the community of individuals with an autism diagnosis is so much larger than the community of individuals with fragile X, there are many additional therapies, treatments, and interventions that can be very relevant for our kids. For example, we've found that applied behavior analysis (ABA) and applied verbal behavior interventions have been very effective educational approaches for our son. I strongly encourage any parents of a child with a dual diagnosis (autism and fragile X syndrome) to explore and learn about the many autism-related resources available to them. A good place to start is the Autism Society of America web site (www.autism-society.org).

Steve McKee
(father and NFXF Board Member)

Children with FXS who were not autistic had a behavioral profile that was similar to the controls with developmental disabilities. In addition, the group with both FXS and autism were the lowest of all the groups on developmental testing, results similar to the studies of Bailey and colleagues.

The reason why some children with FXS have autism too, may relate to additional background gene effects (other genes inherited from their mother and father) and further studies are underway. There also are effects of the fragile X protein deficit that predispose children to autism including the hyperarousal to stimuli and the shyness and social anxiety. When these problems are severe in FXS, then autism is more likely to occur.

A recent report by Roberts et al. 2002 demonstrated more autonomic dysfunction (problems with the sympathetic or parasympathetic nervous system such as increased heart rate when scared or stressed) and hyperarousal in children with both FXS and autism compared to FXS alone. Also of note is a recent neuroimaging study in females which demonstrated that the size of the posterior cerebellar vermis (an area of the brain involved in motor function, cognition and sensory perception) on MRI correlated inversely with the number of autistic features. In addition, the number of autistic feature also correlated with the severity of anxiety (Mazzocco, Kates et al. 1997).

More research is needed regarding treatment of children with both FXS and autism. Preliminary studies in autism suggest that early treatment with a selective serotonin reuptake inhibitor (medications like Prozac,

Paxil, Luvox and others) is beneficial for both language and socialization skills (DeLong, Teague et al. 1998) and this possibility needs to be studied in children with FXS and autism. Intensive behavioral interventions are helpful in young children with autism and this too needs to be evaluated in young children with FXS and autism together (Scharfenaker, O'Connor et al. 2002).

Finally, it is important to identify the children with FXS among those who have been diagnosed with autism alone to provide genetic counseling and access to treatments and interventions known to be beneficial for individuals with FXS. Previous screening studies have shown that 2.5% to 6% of boys with autism have FXS. (Brown, Jenkins et al. 1986; Bailey, Phillips et al. 1996; Hagerman 2002) Therefore all children with autism and or mental retardation should have fragile X DNA testing. Such screening may also identify individuals with the fragile X premutation in association with autism and we are currently evaluating the additive effect of the premutation which can be associated with mild gene dysfunction (Tassone, Hagerman et al. 2000).

In summary, the association of fragile X syndrome and autism is a strong association which requires assessment in each child and will guide future treatment endeavors.

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A Fab Four Fundraiser for Fragile X!

On April 6, 2002, the Fragile X Resource Group of Greater Chicago led friends and family on a trip down memory lane (or should we say “Penny Lane”?) when they hosted a fab-



Paul, George, Ringo, and John, and fans at the “Party at the Park” fundraiser. This “Fab Four”—the Beatles tribute band American English—helped raise over \$55,000 for the Foundation.

ulous evening in support of the National Fragile X Foundation. Over 300 people attended the “Party at the Park” enjoying cocktails, dinner, a silent auction, dancing and a concert by internationally acclaimed Beatles tribute band, *American English* at the trendy Park West theatre in Chicago.

The event was a great success, netting over \$55,000 in support of the Foundation’s important work—promoting awareness, supporting research and aiding families affected by fragile X syndrome. In addition to those who attended, many others who were not able to attend sent in donations.

We all “get by with a little help from our friends.” A special thanks to all of our family and friends for their love, understanding and support!

Lynda Canel
Avis Primack and
Maureen Schmidgall
2002 “Party at the Park” Co-Chairs

FRAXA Update

In February and March, FRAXA funded thirteen research projects at universities in the United States and Canada. The projects are described in the latest issue of our newsletter, which is available upon request. Probably the most exciting of these studies is a clinical trial of a drug which may help people with fragile X learn.

Over the past decade, several pharmaceutical companies have been developing a new class of promising medications, called “Ampakines”, which seem to enhance learning. FRAXA’s very first 1994 newsletter featured an article by Dr. Michael Tranfaglia (psychiatrist, FRAXA co-founder, and also a fragile X parent) about the potential that Ampakines offered for people with Fragile X. For the past year, FRAXA has been funding a study of one such compound in the Fragile X knockout mice, in the laboratory of Dr. William Greenough. Now FRAXA has approved \$72,000 to fund a trial of this compound in adults with Fragile X, under the direction of Dr. Elizabeth Berry-Kravis, at RUSH University in Chicago. Dr. Berry-Kravis

states “this treatment trial with CX516 represents the first ever attempt at treatment directly aimed at reversing the intellectual disability in Fragile X.”

If you would like to learn more about this study or other projects FRAXA is supporting, visit www.fraxa.org or contact me at:

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Editor’s Note: 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.

Responding To the Death Of A Teenage Girl with Fragile X Syndrome

A NFXF fragile X resource center helps in a crisis

Margaret Israel
President,
Fragile X Resource Center of Missouri

It was supposed to be a Valentine's Day party but it ended with the death of a sixteen year old girl with Fragile X Syndrome (FXS). When the students were leaving their classroom to attend the party in another room, the young girl was told not to bring the soda can she was holding, and she began throwing a tantrum. Apparently the tantrum escalated to the point that teachers felt she was uncontrollable and she was moved to a crisis intervention room. Several teachers held her down by her arms and legs. She suddenly stopped breathing. Police and emergency medical personnel were called but it was to no avail. The teenager died in that room. The results of the autopsy were inconclusive, with no apparent physiological cause of death determined. The St. Louis County Medical Examiner ruled that the student died of "agitated delirium", a rare cause of sudden death that cannot be verified by autopsy. What is known is that just 24 hours earlier she was feeling fine and having a great time at a school dance.

When I read the St. Louis Post-Dispatch newspaper account that "she suffered from fragile X syndrome", I was immediately concerned that readers would assume fragile X is a fatal condition. I was also concerned that despite our local group's efforts to provide information to the general community, and specifically to the educators who work with our children, most people still had a minimal awareness of FXS or of appropriate and effective interventions that perhaps might have prevented this catastrophe.

Upon learning of the fragile X connection, I spoke with Robby Miller at the National Fragile X Foundation to inform him of the situation and of my intention to speak with the Special School District administrators. He fully agreed with the need to work with the District and to provide accurate information about fragile X.

I have been very involved with the Special School District over the years, sitting on numerous committees and serving as President of the Citizens Advisory Council. Our Fragile X group meetings take place at the district's administrative building. As soon as I learned of this tragic event I called the Superintendent and the Assistant Superintendent for Curriculum Development. My purpose was not to assess blame, but rather to use this tragic event as an opportunity to provide teachers with information about FXS and to suggest techniques for working with students who have difficulty with transitions, have high levels of anxiety, and have expressive language delays.

Initially I did not know the identity of this young girl. But within a short time her mother telephoned and we had a poignant conversation about how the family was dealing with their sudden and untimely loss. The family had previously requested information from our Resource Center, received all our mailings, and attended one of our fragile X family picnics. The mother was very supportive of the plan to conduct a workshop on FXS to enlighten educators and parents. We also agreed that it was an appropriate way to honor her daughter's memory.

I met with the Superintendent to suggest that our Resource Center and the District co-sponsor a symposium on fragile X syndrome. I suggested Marcia Braden, Ph.D. as a keynote speaker because of her vast experience working with individuals with FXS.

With the financial support of the National Fragile X Foundation, we arranged for Dr. Braden to come to St. Louis. All of our local families were informed of the incident and invited to assist in the positive steps we were taking to address this serious and sad turn of events. Everyone involved understood that it was critical to present accurate information about characteristics of fragile X and approaches to addressing behaviors and learning styles.

The symposium was held on February 4–5 2002, almost a year after the student's death. The first day-long workshop titled *Fragile X Syndrome Through The Life Span* gave an overview of Fragile X as well as a comparison/contrast among autism, Aspergers and FXS. The evening session, *Techniques in Addressing Behaviors*, was an informal session designed for parents.

The next day's session addressed the heart of what we wanted the special education staff to understand and implement as they worked in their classrooms. The session was entitled *Programming Issues for Fragile X and Related Disorders: Functional Assessment and Development of Effective Behavioral Support Plans* and provided an opportunity for teachers to present specific cases and issues and receive valuable tips for effective classroom interactions. All sessions were well attended and well received by teachers, therapists and parents alike.

The Special School District of St. Louis County and our Fragile X Resource Center of Missouri worked together to turn a tragic death into a positive learning experience. We may never know why our sixteen-year-old friend died on her way to celebrate Valentine's Day, but she remains in our hearts. She is an inspiration to continue research on fragile X syndrome and to continue to provide critically necessary information on FXS to our parents, our teachers, and the general public.

Reserve Now! The most colorful event of the year!

During the upcoming 8th International Fragile X Conference there is an optional dinner cruise on Friday evening. The "duck" will provide a moonlit cruise on Chicago's Lake Michigan.



The cruise has something for everyone including a newly-constructed, 500-passenger vessel built specifically for cruising the Chicago skyline on three enclosed decks plus an open-air deck to explore!...incredible skyline views!...largest picture windows of any ship at Navy Pier. A great view is available from every seat, but if you still crave a more "wide-open" perspective, just walk on up to the open-air flight deck!

A bountiful buffet—great food and lots of it, with enough variety to satisfy anyone's taste! That's the duck buffet where you'll find fresh, creative salads, entrees and desserts, prepared fresh on board, daily, plus complimentary coffee, tea and water.

Fun entertainment—the emcee will keep everyone entertained while four decades of great music beckon you to the two huge dance floors.

Great service—friendly, courteous, and fun. That's the professional duck crew. Everyone from the reservations staff to the serving and marine crews are ready to assist you. They strive to make every cruise a great one!

See page 27 of this newsletter to register for the conference and sign up for the dinner cruise. If you have already registered for the conference, and wish to add the dinner cruise to your registration, no problem!

Just call the NFXF at 1-800-688-8765 to add the cruise to your plans.

Parent's Forum



EDITOR'S NOTE: A letter and a copy of an article arrived at the National Fragile X Foundation from Mr. Dale Thompson, Jr. It detailed a day in the life of a single father raising a son diagnosed with fragile X syndrome. The article had been published in the Wilmington Star on June 25, 2001, and described the challenges and love between Dale and Johnathon. I loved the story and asked Mr. Thompson to write his feelings about his life as a single father for us.

- Cindi Rogers, CIJRogers@aol.com

A Dad's Story

Dale Thompson, Jr.

I remember the day Johnathon was born, October 16, 1997, at 2:05 p.m. My life was changed forever! There was my beautiful 8 lb. 2 oz, 20" long little baby boy. My heart instantly belonged to him.

Johnathon's mother and I were separated at the time of his birth, and reconciled for only one month after. This was a very difficult time for me. I wanted to be with my son so bad. When Johnathon was thirteen months old, his doctor, Belinda McPherson, who had been his doctor since he was three days old, noticed he was not developing as he should. She referred him to Duke Medical Center in Chapel Hill, N.C., where he was diagnosed with fragile X syndrome. I was devastated!

I had been fighting for custody of Johnathon since he was about six months old due to the fact that he was not being cared for properly. Being



diagnosed with fragile X only made me want him more because he would need a lot of love and care. Finally, on September 10, 1999, thanks to my great attorney, Laura Thompson (no relation), of Southport, N.C., I gained custody of my son. What a great day in my life! Johnathon was now going to get the love and care he deserved.

Exactly one week later we lost almost everything when Hurricane Floyd washed through our home and devastated the area. Johnathon and I have worked together to rebuild what Floyd destroyed. It didn't destroy our determination!

A month or so before this time, Johnathon began attending a special school at the Child

Development Center (CDC) in Wilmington, N.C. where he still goes today. I drive approximately 100 miles a day to get him there and back. He receives speech therapy to help him talk, OT to help him dress himself, use his hands and develop body strength, and many other things (too many to list).

Recently, Johnathon was tested at the Developmental Evaluation Center in Wilmington, N.C. for admission into the Kindergarten program. He was found to be moderately impaired. Johnathon has gone above and beyond all of my expectations of him.

Being a single parent is tough, but I would not have it any other way. I like being the Mom and Dad. I have many people to thank for helping Johnathon and me. The National Fragile X Foundation has helped us with all of the wonderful work they do; Dr. McPherson for recognizing Johnathon's delays and having the courage to send him to Duke; my attorney, Laura Thompson for helping me gain the love of my life; the CDC for helping Johnathon learn the things that will help him be a part of life and the community; and most of all my friends, family and Mount Holly Baptist Church. Without all of these supports, I could not be the Dad that I am.

Johnathon is a sweet little boy. To know him is to love him. I am very proud of him. Now, if I could just get him to stop calling me "Mommy", and just call me "Daddy".

In the Market for Long Term Care Insurance?

Support the National Fragile X Foundation and Save 10% Yearly!

Long Term Care Insurance (LTCi) is one of the most rapidly expanding markets for insurance. As the baby boom generation approaches middle age and even retirement many are considering this coverage which can pay for skilled or custodial care at home or in a nursing facility. A friend of the National Fragile X Foundation has recently helped to establish an affinity arrangement with CNA Insurance. Through this arrangement members of the National Fragile X Foundation at the Basic Level and above (\$25.00 or more yearly) qualify for a 10% yearly premium discount on Long Term Care Insurance. We have just processed our first affinity membership and after an initial donation to the NFXF of \$300.00 our newest member is saving over \$600.00 yearly on LTCi.

We're not in the insurance business, we receive nothing from CNA and we're not recommending Long Term Care Insurance or CNA. But if you're in the market, a yearly tax deductible donation to the NFXF could save you premium dollars and help support the mission of the NFXF*. As always, we recommend that you rely on trusted financial advisors, insurance agents and accountants when making these important decisions.

**Please contact the NFXF for a CNA affinity membership form that will entitle you to a 10% annual discount if you apply to and are accepted by CNA Insurance. You must obtain this form prior to applying for coverage.*

Announcing the new and 3rd edition of

Fragile X Syndrome: Diagnosis, Treatment, and Research

Edited by Drs. Randi and Paul Hagerman, released by Johns Hopkins University Press

—Now available from the National Fragile X Foundation

Substantially revised and updated, this book discusses the clinical approach to diagnosing the disorder, supported by the latest research in epidemiology, molecular biology and genetics, and neuropsychology. It also presents information on treatment: genetic counseling, pharmacotherapy, intervention, and gene therapy.

Praise for previous editions:

“Includes updated chapters on the cytogenetic and molecular biology of the FXS mutation and premutation. The first half addresses the diagnosis and research aspects and is well referenced. The latter half is dedicated to treatment and intervention. The chapter that emphasizes an integrated approach to intervention could easily qualify for continuing medical education credit.”

— *Journal of the American Medical Association*

“This book should sit on the library shelves of clinical geneticists. It is well written, well referenced, and should become well thumbed.”

— *Journal of Medical Genetics*

“Answers nearly all the questions that parents or clinicians might raise about fragile X syndrome . . . Can be recommended confidently as a thoroughly up-to-date, reliable, and informative account of the condition.”

— *Lancet*

“The clinical and cytogenetic material in this book is excellent and provides a strong background for physicians and students... *Fragile X Syndrome* still presents the best comprehensive treatment of this complex disorder. Physicians, students, and other interested professionals can either read this book from cover to cover or select the chapters that interest or apply to them.”

— *New England Journal of Medicine*

Randi Janssen Hagerman, M.D., is Tsakopoulos-Vismara Professor of Pediatrics at the M.I.N.D. Institute in Sacramento, California. Paul J. Hagerman, M.D., Ph.D., is a professor in the Department of Biological Chemistry at the University of California, Davis.

EDITOR'S NOTE: The prior, and 2nd edition of this comprehensive medical guide was released in 1996. We are pleased and excited to see this new volume made available. Please keep in mind, however, that this book, while filled with important information for families as well as professionals, is technical in nature and can be difficult reading for those unaccustomed to books of this nature. This book is now available from the NFXF. Please see the Order Form for National Fragile X Foundation Resources in the back of this issue.

NFXF Education Project

Carolyn Ybarra, Ph.D, Project Coordinator

A Practical Resource for Teachers— *A project in honor of student Brett Dobslaw*

Project Update:

The National Fragile X Foundation's 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 a published binder of information.

We are off to a good start, with a project team including therapists, teachers, and parents. We have an outline for the binder, and writing has begun on the general sections that describe Fragile X and give guidelines for adapting curricula. The lesson planning section for teachers will include ideas and examples for reading, writing, communication skills, math, and vocational studies, among others. We are considering having pull-out sheets for each category for teachers to use with students at various levels of ability.

The Education Project Team is planning two conference sessions at the 8th International Fragile X Conference in July. During the Education Project Workshop, tentatively scheduled for Thursday, July 18, at 9:45 a.m., participants from the Education Project Team will introduce and describe project plans. We also will provide a forum for input from workshop attendees. Please attend and bring your brainstorming ideas about approaches to academics, particularly reading and math, which have worked well for children with fragile X.

The Master Teachers Panel, tentatively scheduled for Friday, July 19, at 3:05 p.m., will feature four experienced teachers who will discuss or demonstrate lesson plans and activities that have been successfully used to educate children with fragile X. We were happy to have received nominations from all over the country of people who have experience teaching our children. While the Master Teachers panelists have already been chosen, we continue to accept input from experienced teachers who can advise our project team. If you know of a successful teacher of children with fragile X, please let us know.

Please also let us know if you have used, or seen used, software, materials, classroom or lesson adaptations, or other techniques that have worked particularly well with your child. You can email me at ybarra@stanford.edu, or fax me at 650-367-7571. Postal mail can be sent to me c/o the NFXF, Education Project, P.O. Box 190488, San Francisco, CA 94119. Feel free to send copies of material, references, or just a description, along with your contact information.

Carolyn Ybarra, Ph.D, Project Coordinator
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EDITOR'S NOTE: This project is made possible through the generous support of the Dobslaw family of New Jersey.

Celebrate National Fragile X Awareness Day July 22

Call the medical reporter at your local newspaper and TV stations and ask them to acknowledge National Fragile X Awareness Day.
If they're interested call our office and we'll send a press packet to them.

NATIONAL FRAGILE X FOUNDATION

UPDATED: 4-19-02

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Florida: Conquer Fragile X Foundation
Parent Support Network - affiliated with The
National Fragile X Foundation

For the following South Florida Counties only:

Dade, Broward, Palm Beach and Martin
Contact: Adrienne Griffin
Tel: 561-842-9219
Fax: 561-842-0597
E-Mail: mail@cfxf.org
Internet: www.conquerfragileX.org

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(Lisa Scott, genetic counselor)
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E-Mail: lscott@genetics.emory.edu
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ALLIANCE OF INTERNATIONAL FRAGILE X PARENT SUPPORT GROUPS

Updated 04-18-02. Please note: As administrator for the Alliance, the National Fragile X Foundation attempts to maintain a current and accurate list of international parent support groups. However, the NFXF cannot guarantee the accuracy of all information. Please visit www.FragileX.org for regular updates to this list.

Argentina

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Email: enrichd@tpg.com.au
Web: <http://members.ozemail.com.au/~fragilex/>

Queensland Fragile X Association
Email: d.birney@uq.net.au

Belgium

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Email: mbaetens@belgacom.net
Web: <http://titan.glo.be/janj/fragiel.html>
Web: <http://users.pandora.be/chevalier/onder.htm>

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(Also includes support for fragile X families)
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Email: info@fragilex.k-web.co.uk
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Uruguay

Asociacion X Fragil del Uruguay
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Politics and such...

John Harrigan, Public Policy Chair

The FDA recently backed off from a proposal to suspend a rule requiring drug companies to perform safety testing of adult medicines commonly given to children. The agency had, according to the AP, attempted to take the position that because of the recently enacted BPCA legislation—which was supported by the NFXF and the Coalition for Children's Health—suspension of the rule mandating pediatric testing would not be harmful as it was otherwise required in the recent legislation. The NFXF and the CCH, along with other groups, registered their protests and the FDA relented when faced with mounting protests and additional legislative action... the NIH reports that, in 2001, five of its Institutes spent \$11.7 million in funding for FXS research. Appropriations language out of the House and Senate will, hopefully, result in a substantial increase in that commitment next fiscal year. Kudos to David Busby for his usual

yeoman's work in this area and thanks to the many NFXF members who have written and/or talked with their members of Congress and aides on these issues. The CCH supports funding level increases for all programs with regard to fragile X to increase to \$33 million next fiscal year. We are pleased to announce that Dr. Mary Lou Oster-Granite of the NICHD at NIH will join our Public Policy panel at the upcoming Chicago conference... NFXF is also currently interested in and tracking IDEA Re-authorization, Genetic Nondiscrimination in Health Insurance and Employment legislation, amendments to the Orphan Drug Grant Program, and expansion of the Office for Rare Diseases at NIH. We are working individually and in coalition with CCH and NORD (National Organization of Rare Diseases). Contact NFXF if you have interest in any of these matters.

EDITOR'S NOTE: In the Winter 2002 issue of the Foundation Quarterly, the National Fragile X Foundation highlighted the wonderful accomplishments of Jonathan Doring from Florida. Jonathan is an exceptional young man who deserves our admiration. However, we believe it important that our readers be aware that Jonathan does not have fragile X syndrome (fraxa), but rather fraxe, a milder X-linked disorder, and much more rare than fragile X syndrome. This fact was unknown to us at the time of publication. To better understand fraxe, please read the following article from one of the members of our Scientific and Clinical Advisory Committee. Jonathan will also be speaking at our International Conference as part of a special session on fraxe.

A Tale of Two Fragile X Sites FRAXA and FRAXE

David Nelson, Ph.D.

Baylor College of Medicine, Dept. of Molecular Genetics

In the days shortly after identification of the CGG repeat expansion in the *FMR1* gene, it became clear that what we knew as fragile X syndrome was really two different disorders caused by defects in two different genes, each affected by CGG repeats. It turned out that some of the families that were being studied as having fragile X syndrome did not show the repeat expansion in the *FMR1* gene, in spite of having a fragile site on their X chromosomes in what was apparently the same place. Laboratories quickly determined that these families had a fragile site that was a short distance away from the one in the *FMR1* gene, and this newly recognized fragile site became known as *FRAXE* (*FRAXA* being the first fragile site on the X chromosome, and *FRAXE* being the 5th to have been found).

As families were sorted into having either *FRAXA* or *FRAXE*, it became clear that *FRAXE* disease was rather rare (maybe 1/100 as common as *FRAXA*), and that the disease was somewhat milder, with few, if any, of the physical features found in patients with *FRAXA*. Yet the repeat expansions were

very similar. Both involved CGG repeats that became very large in affected individuals.

It took a few more years before the gene affected by repeat expansion at *FRAXE* was characterized. This gene is now known as *FMR2*. It is not at all like *FMR1*. It makes a completely different protein that is found very early in fetal development and at low levels in the brains of adults. The *FMR2* protein is found in the nuclei of cells, and appears to be involved in regulating the activity of genes, possibly working to help cells determine what type of cell they will become. Less is known about the *FMR2/FRAXE* gene and its protein than those of *FMR1/FRAXA*, although recent advances have begun to shed light on *FMR2*'s normal function and the consequences of its absence in individuals with *FRAXE*. Very recently, a mouse model for *FRAXE* was developed, and this should provide significant new data about the disease in humans.

FRAXE families have been involved in fragile X research and in the various foundations dedicated to support of families and individuals with fragile X syndrome since the beginning of these efforts. Although their form of the fragile X syndrome is somewhat different, they are no less welcome in our community. They have a great deal to contribute to both our scientific and social understanding of developmental disability, and suffer from a very similar (and nearby!) disorder.

NFXF Research Fund

Fellowship Recipients Report on their Efforts

Observation of Social Anxiety in Children with Fragile X Syndrome

Pearl Chang

2001 LeCover Memorial Fund Recipient

To better understand the genetic and environmental factors that influence the development of children with fragile X syndrome, last summer I had the unique opportunity to observe children affected by this syndrome. In the past 2 years, Dr. Allan Reiss' team, part of the Stanford Division of Child Psychiatry, visited 120 families who had children with fragile X. The in-home visit included a videotaped "social challenge" task, which both the children with fragile X and their unaffected siblings participated in. The social challenge was designed to measure and evaluate how children with fragile X respond to a new social situation and their level of stress and anxiety compared to their unaffected siblings. Dr. Reiss and his team were particularly interested in children affected by fragile X because they are known to have elevated levels of social anxiety, and it is important to learn how these children respond in social interactions as well as what factors influence their response.

Under Dr. Hessel's supervision, another Stanford student, Milena Petranovic, and I analyzed the videotapes of the social challenge, which included an interview, a silent reading, an oral reading, and singing a song. We used a standardized rating system to assess specific behaviors, including gaze, vocal quality, speech length, discomfort, and avoidance of task. In our observations, we learned to detect both the outright displays of anxiety, as well as what may be subtle signs of discomfort, such as nervous laughter or frequent facial tics.

Because of the structured nature of the social challenge and the currently known symptoms of children with fragile X, I initially assumed that all the videotapes would turn out to be more or less the same. I expected children with fragile X to respond adversely to the social challenge, while their siblings would complete the tasks well. However, this was not the clear-

Continued, page 25

Mathematical Skill Development Among Girls With Fragile X

Gwyn Gerner

2001 Rosen Research Fund Recipient

The research study in which I participated allowed me to expand my understanding of fragile X syndrome, specifically in the area of mathematical skill development among girls with fragile X syndrome. This study, which was supported by the Rosen Research Fund is an extension of the current research being completed by my research mentor, Michele Mazzocco, Ph.D. for the Math Skills Development Project at Kennedy Krieger Institute. This ongoing research is being conducted in order to identify whether weak math achievement is observed in girls with fragile X syndrome, as well as other populations during primary school age years, and whether there are any specific components of such early math weakness. As a part-time research assistant in the Math Skills Development Project, I have been predominately involved in research on math achievement in children with Neurofibromatosis type 1 and research being conducted in order to identify weak math achievement or math disability in elementary school children. This was my first opportunity to participate in this specific area of research on fragile X syndrome. This opportunity allowed me to expand my research skills in areas such as research methodology, statistical analyses, and item analyses, which complemented and added to my current graduate work.

Throughout this project supported by the Rosen Research Fund, I have been involved in the coding of cognitive data, including data analyses procedures and methods. In addition, I have been involved in some manuscript preparation activities. The focus of this project is the examination of the specific types of conceptual or procedural categories of math items that are particularly difficult for young girls with fragile X, relative to a paired comparison group. The results of this work will be submitted for publication after additional participants are recruited. Although the findings of this

Continued, page 24

Brain Structure and Connectivity in Neurogenetic Disorders through Neuroimaging

Marie Holzapfel

2001 Rosen Research Fund Recipient

It was my privilege to receive the Rosen Research Fund Summer Fellowship for investigation of the brain's biological components and how they correspond to the behavior and cognition of patients with fragile X syndrome. As a research assistant for Dr. Allan Reiss, Director of the Behavioral Neurogenetics Research Center at the Stanford University School of Medicine, my work involves elucidating brain structure and connectivity in neurogenetic disorders through neuroimaging. Neuroimaging offers great potential to expand the knowledge of these disorders through structural visualizations of the living human brain. Ultimately, the biological markers it provides help in guiding research into new interventions and treatments.

Neuroimaging studies of patients with fragile X syndrome indicate that the FMRI gene mutation affects development, growth, and maintenance of the brain. While the brain consists of both gray and white matter, abnormalities in the white matter circuits could help explain attention dysfunction, hyperactivity, executive function deficits, and impulse control characteristics of fragile X syndrome. To measure this white matter connectivity, I used a new and innovative magnetic resonance imaging method, called diffusion tensor imaging (DTI).

Working with Dr. Naama Barnea-Goraly, a post-doctoral research fellow in the lab, I used DTI to examine white matter structure and locate the pathways involved in cognitive and behavioral abnormalities associated with fragile X syndrome. Using DTI brain scans from individuals with fragile X and healthy controls, my role was to analyze the data using region of interest (ROI) analysis. Each ROI was digitally drawn on each subject's scan to include only white matter within the specified regions of the brain. The analysis of these data is currently underway.

I would like to express my appreciation for the

Continued, page 24

Discourse and Conversational Aspects of Females with Fragile X

Katarzyna Lesniak-Karpiak

2001 LeCover Memorial Fund Recipient

The LeCover Memorial Fund award allowed me to expand my research interest in fragile X syndrome and specifically to partake in a project on discourse and conversational aspects of females with fragile X. My research is a continuation of earlier work that I carried out under the supervision of my research mentor, Michele Mazzocco, Ph.D., when I was completing my clinical psychology internship at Kennedy Krieger Institute/Johns Hopkins Hospital. As an intern I volunteered to participate in a study of social behaviors in girls with fragile X syndrome. Working with Dr. Mazzocco, I examined behaviors of children with fragile X syndrome during initial anxiety provoking social situations. I viewed videotapes of participants who were to engage in conversation with an unfamiliar adult. My participation and involvement in this project entailed coding data, analyzing the data, and subsequently writing up the results. The main findings of the project indicted that the females with fragile X took longer to initiate speech and made more hand movements than the other participant groups, but the total time engaged in the conversation was not different among groups. Contrary to predications, there were no significant differences in other anxiety related behavioral markers including the frequency of eye gaze, rigidity or fidgeting, or facial movements. Our manuscript was submitted for publication in March 2001 and will be published in 2002.

In the process of my involvement with the project described above, I remained blind to the group membership of the participants. The characteristics of discourse I observed during the social interactions peaked my interest and desire to examine the nature of conversational characteristics of females with fragile X. Although impairments in speech and language of males with fragile X are well documented, research on language and discourse quality of females with fragile X

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NFXF Research Fund

Fellowship Recipients Report on their Effort (continued)

Predictive test for cognitive functioning in female carriers of fragile X syndrome using hair root analysis

Sandra van't Padje, *Dept of Clinical Genetics, Erasmus University Rotterdam*

2001 NFXF Research Fund Recipient

To investigate the molecular basis of the spectrum of cognitive functioning in female FM carriers and to evaluate the predictive power of the FMRP assay on hair roots, I have compared FMRP expression in hair roots and basic cognitive functioning in female FM carriers. In first instance I started to learn the practical aspects of the FMRP test by analyzing FMRP expression in hair roots from control individuals (n=25). Myself performed the plucking and all the steps of the immunoincubation. During this training period Dr. Rob Willemsen and technicians in his lab supervised my results. Also the examination and scoring of the hair roots were performed independently using microscopy. After obtaining sufficient practical experience with these control samples, I continued my study using hair roots from female FM carriers whose CGG repeat size had already been tested by DNA analysis. In total, hair roots from 10 female FM carriers were analyzed for FMRP expression. For this purpose, I visited the females at home to pluck hairs and under supervision of staff members of the Dept of Orthopaedagogics all participants were individually administered the Standard Raven's Progressive Matrices during this home visit.

The next day I started to analyze the hair roots from female FM carriers in the lab (in a blind fashion) for FMRP expression. In the same analysis we included control samples (also blind). The analysis of the Raven test was performed in the Dept. of Orthopaedagogics. The results of my experiments were included in the statistical analysis of a large group of female FM carriers who were tested for FMRP expression in hair roots and mental status (n>40).

From this large study-group we found evidence that female FM carriers with normal intelligence had a normal percentage of hair roots that expressed FMRP,

whereas FM carriers with mental impairment had a reduced percentage of hair roots that expressed FMRP. We conclude that the FMRP test on hair roots is a strong prognostic indicator of cognitive functioning in female FM carriers and has great potential as a predictive test.

Finally, I would like to express my thanks to The National Fragile X Foundation's Research Fund for giving me the opportunity to work on this topic as a summer student. During this period my motivation to work on research linked to fragile X was further increased and I have decided to accept a Ph.D.-student position on fragile X research in Rotterdam.

Gwyn Gerner, *continued from page 22*

research were consistent with the research suggesting that girls with fragile X have a higher rate of poor math achievement relative to a paired comparison group (Mazzocco & Myers, 2000), the specific nature of the deficits were somewhat inconclusive. Therefore, additional participants will be recruited to obtain data to clarify the current findings.

The generosity of the Rosen Research Fund has been instrumental in expanding my role in the current fragile X research being conducted at the Math Skills Development Project. It is hoped that the current research will make a scientific contribution and initiate further research in this area. I would like to extend my gratitude to the National Fragile X Foundation Rosen Research Fund for the opportunity to actively assist in conducting this research and participating in this valuable learning experience.

Marie Holzapfel, *continued from page 23*

Rosen Research Fund Summer Fellowship award. Not only has this award allowed me to develop my personal interests in neuroscience, but it has given me the invaluable opportunity to participate in the effort to further understand the complexity of fragile X syndrome.

Pearl Chang, *continued from page 22*

cut picture that I encountered. Many children with fragile X performed as well, or even better, than their siblings, and appeared to enjoy the challenge. They were poised as well as enthusiastic, and it was only afterwards that I realized their diagnosis. In contrast, there were also unaffected siblings who seemed to dread the social challenge and appeared nervous and tense.

I was also surprised by the wide range of behaviors exhibited in both children with fragile X and their siblings in response to the social challenge. While I didn't think children with fragile X could handle the task of singing a song, many responded well. A few children seemed to embrace the task—one boy even performed a full impersonation of Elvis. At the same time, while I expected the silent and oral readings to be fairly low-anxiety level tasks, many children with fragile X could not complete the task and a few siblings had trouble as well.

Anxiety, especially social anxiety, is something that affects everyone, perhaps more so in some than in others. Therefore, being able to cope with anxiety or stress is an important life skill. Data from the social challenge coding is currently being analyzed, and hopefully it will provide some insight into what aspects of the social challenge and social situations in general elicit the most anxiety-related behaviors in children with fragile X and how and why they respond similarly or differently. Then, perhaps future medical and behavioral interventions can be developed to help children with fragile X overcome social anxiety if needed.

I would like to thank the National Fragile X Foundation, the Harry LeCover Memorial Fund, Dr. Reiss, Dr. Hessel, and the Stanford Division of Child Psychiatry for providing me with this unique opportunity not only to learn about fragile X syndrome, but to observe children with fragile X and be part of the ongoing research effort to understand the problems that individuals with fragile X face and what can be done to help them. I hope that my efforts will contribute to a better understanding of fragile X syndrome and ultimately be a benefit to the fragile X community.

Katarzyna Lesniak-Karpiak, *continued from page 23*

syndrome is sparse. Furthermore, the underlying reasons for the observed language deficits are also not fully understood. Abnormal regulation of arousal, social anxiety, or self-monitoring deficits have been suggested as potentially contributory factors to the language problems observed in males with fragile X syndrome. To provide preliminary information on the discourse quality of females with fragile X, in the project supported by the LeCover Memorial Fund, I began studying the nature of social discourse using transcriptions of videotapes described above. It is hoped that this study will help us to better understand the quality of discourse patterns and the relationship between emotional or cognitive aspects and the communication skills of females with fragile X syndrome.

After receiving the LeCover award, I was involved in revising the manuscript assessing behavioral markers of social anxiety in females with fragile X. In addition, I reviewed the literature on discourse characteristics in fragile X and assisted in operationalizing discourse criteria that are used to evaluate the quality of language

of the participants for the current study. I also participated in training to code language usage at the utterance level. Of particular interest is our examination of atypical aspects of utterances such as dysfluencies and repetitions. The manuscript resulting from this work will be submitted for a publication in collaboration with Dr. Mazzocco and other collaborators that I met through this fellowship experience. Without the generous support of the LeCover Fund this project would not have been initiated. I appreciate the opportunity to spearhead this important project. I also hope that this study will promote interest and additional research projects on this topic.

...this study will help... understand the quality of discourse patterns and the relationship between emotional or cognitive aspects and the communication skills of females with fragile X syndrome.

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

Sheraton Chicago Hotel & Towers, Chicago, Illinois USA

CONFERENCE REGISTRATION FEE SCHEDULE

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

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

Type	Regular Registration 5/18/02-6/30/02
Professionals	\$415
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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.

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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.)

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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)

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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.

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**. _____

TOTAL PAYMENT Please include meal ticket and dinner cruise fees, plus registration fees calculated from Conference

Registration Fee Schedule, and any additional fees (Childcare fees are payable at the door.): **Total amount due \$** _____

Payment in full must be received by June 30, 2002. Cancellations through June 30, 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 _____

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

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FXF Welcomes Gene Warren to the Board of Directors!

In Gene's own words...

As the Chief Operating Officer at ACT Teleconferencing in Golden, Colorado, I am responsible for managing the worldwide sales activity and service delivery of an \$85 million organization.



I have over 25 years of executive and technical experience in the telecommunications field. But, more importantly, I am a husband and proud father of four incredible children who bring joy to my life and challenge me in such unique ways.

At the age of 3, my son Chris was diagnosed with fragile X syndrome. For the last 19 years, fragile X has been part of our family and I have made it a lifetime commitment to work with the National Fragile X Foundation. It is my desire to serve on the board of directors because there are a lot of kids that need help. There are also a lot of parents and families that need support as well. I want to be there to make a difference.

When I was growing up, I was one that needed help, and now in return, I have the opportunity to contribute to something that is extremely important. As a father of a child with fragile X, and in my position as an executive with business contacts around the globe, I believe I am in the perfect spot to bridge these communities.

It is an honor to become a board member of the National Fragile X Foundation. Thank you for the opportunity to work with all of you.

Sincerely,
Gene Warren

...From the Executive Director

Continued from page 5

Speaking of our conference, the **8th International Fragile X Conference** in Chicago is now organized and ready for primetime. The Preliminary Program Agenda has been online since March (visit www.FragileX.org and click on the red Chicago logo) with changes color-coded to assist with your planning.

Though it seems readily apparent to those of you who have previously attended, it bears repeating that this event is truly life-altering. I say this without hesitation. I have heard this hundreds of times and it was obvious at the last conference in Los Angeles. The "buzz" the excitement, the learning that took place, the friendships that were made, the sharing, the synergy that occurred and the plain ol' fun everyone has, makes for an unforgettable experience.

Last, but not least, it is our great pleasure to announce the availability of the new, and 3rd edition of **Fragile X Syndrome: Diagnosis, Treatment & Research**, edited by Drs. Randi and Paul Hagerman. It has been six years since the 2nd edition and it goes without saying that there has been great progress in understanding and treating fragile X in that time. Please keep in mind, however, that that this book is primarily written for medical, therapeutic, clinical and educational professionals. It can be slow-going for even some of us who deal with fragile X on a day-to-day basis. Nonetheless, with those cautions in mind, parents will find it filled with information that will be beneficial to those who work with your children. (Please find our order form in the back of this newsletter.)

I look forward to seeing and meeting many of you in Chicago. In the meantime, please don't hesitate to let us know how the National Fragile X Foundation can be of service to you and your family.

Robby Miller

Conference Reservations
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Genes, School and Family...

Continued from page 4

What conclusions can be drawn from these findings? Most importantly, the results confirm that even among a group of children affected by a specific genetic syndrome, both biological and environmental contributions to development are present. We see that many, though not all, of the family-, school-, and child-based factors that we know to influence typically developing children also affect children with fragile X, despite their level of impairment. Second, noting the role of FMRP, specifying target skills such as attention and computation skills may be most beneficial for measuring effectiveness of biological interventions such as gene therapy and enhancement of neural resources affected by FMRP. Third, the results indicate that children with fragile X and their families may benefit from home-based interventions and support. These interventions may aid the development of cognitive and self-help skills and reduce the severity of behavior problems, in addition to benefits that may be gained from traditional therapies and medication. Fourth, in this study, children receiving educational and therapeutic services appropriate to their needs had less significant behavior problems. However their cognitive and practical skills were not influenced by these services.

Additional work examining the efficacy of particular types of school-based interventions on more specific areas of development such as academic skills is needed. Finally, because this study focused on families at one point in time, it was not possible to determine causal relationships between variables. For example, while we believe that the home environment affects children's development, it may also be the case that having a child or children with developmental disabilities affects the home environment. For many parents the challenges of raising one or more children with a developmental disability may make it very difficult to maintain an environment that is supportive of learning and independence.

Recent funding to revisit these families as well as recruit a limited number of new families has been awarded to Dr. Reiss, making it possible to begin to answer these important causal relationships. If you are interested

in participating in the next phase of this research or would like additional information about the project please contact Dr. Heather Erba at herba@stanford.edu or call (650) 724-3621 or (888) 411-2672.

This work was supported by grants to Dr. Reiss by the National Institute of Mental Health and the Lynda and Scott Canel Fund for Fragile X Research. We greatly appreciate the generous contributions of time and effort made by the many parents and children who participated in this study.

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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

or a history of giving totaling \$5000 or more:

- 50% off on all purchases
- Unlimited copies of The fX Files
- Unlimited FragileX.org Website CDs
- Designate 10 additional recipients of the newsletter
- 5% off on our international conference registration fees

Diamond Membership - \$500 per year:

- 30% off on all purchases
- Unlimited copies of The fX Files
- Unlimited FragileX.org Website CDs
- Designate 6 additional recipients of the newsletter
- 5% off on our international conference registration fees

Supporting Membership - \$100 per year:

- 20% off on all purchases
- Unlimited copies of The fX Files
- Unlimited Fragile X Website CDs
- Designate 4 additional recipients of the newsletter
- 5% off on our international conference registration fees

Family Membership - \$60 per year:

- 10% off on all purchases
- One copy of the fX Files
- One copy of the Fragile X Website CD
- Designate 2 additional recipients of the newsletter
- 5% off on our international conference registration fees

Professional Membership - \$40 per year:

(please indicate below a fragile X related profession.)

- 10% off on all purchases
- One copy The fX Files
- One copy of the Fragile X Website CD
- 5% off on our international conference registration fees

Basic Membership - \$25 per year:

- 10% off on all purchases

* 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

***Designate Additional Newsletter Recipients. Get the word out! *Eligible membership levels entitle you to designate other family members, friends, educators, medical professionals, therapists etc. to receive the Foundation Quarterly. All you do is supply their name and address, and we'll make sure they receive their newsletter.*

Please remember to attach the names and addresses of additional newsletter recipients, if you've selected one of those membership options.

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Name(s): _____

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Please check if

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MEMBERSHIP SELECTION Please check a box below.

Lifetime \$5000

Diamond \$500

Supporting \$100

Family \$60

Professional \$40

Basic \$25

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