

FRAXA UPDATE

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FRAXA RESEARCH
FOUNDATION

“NEVER

DOUBT

that a small

group of

thoughtful,

committed

citizens can

change the

world.

INDEED,

it's the only

thing that

ever has.”

— Margaret Mead

MAJOR RESEARCH ADVANCE

For the first time, scientists have identified specific genes in the brain that are affected by the lack of the fragile X protein. The new research demonstrates that the fragile X protein controls the fate of a number of other proteins in brain cells. This may explain how the absence of this single protein causes the range of physical, cognitive and behavioral symptoms seen in people with fragile X.

Basic research breakthroughs like this one have led us to the knowledge we have today, that will ultimately lead to a cure for fragile X.

Every major discovery so far has increased our understanding of what goes wrong in fragile X and shed light on ways to treat it. This finding is exciting because it links the fragile X protein to thirteen other proteins; these proteins are responsible for normal brain function and for some of the symptoms of fragile X. If we can someday learn how to manipulate these other proteins, this could be another avenue leading us to specific, effective treatments for fragile X.

“Our findings suggest entirely new ways of thinking about treating the problems these patients have,” says Robert B. Darnell, M.D., Ph.D., a principal investigator of the current research. The work is reported in two papers appearing in the Nov. 16 issue of the journal *Cell*. One study, led by Dr. Robert Darnell and Dr. Jennifer Darnell, professors at Rockefeller University, was funded by FRAXA and the National Institutes of Health. The other study was conducted by Steven T. Warren, Ph.D., an investigator at Emory University School of Medicine and Howard Hughes Medical Institute.



Jennifer Darnell, Robert Darnell and Kirk Jensen discover molecular targets of the protein missing from people with fragile X syndrome.

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Does Fragile X Protect Against Cancer?

People with fragile X have a lower risk of cancer than individuals without the disorder, according to Danish researchers. Determining the source of this decreased risk could shed light on how genetic mechanisms prevent cancer from developing.

Also in this issue:

- Report from Washington
- Fragile X Research Centers
- Calendar of Events

The encouraging new findings were reported in the October 2001 issue of the *American Journal of Medical Genetics* (103:226-230). The news was distributed by Reuters News Services to newspapers, television, internet and other news outlets around the world. The fragile X genetic

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FRAXA is a nonprofit, tax-exempt charity run by parents of children with fragile X syndrome. Fragile X syndrome is the most common inherited cause of mental retardation and developmental disabilities, affecting approximately 1 in 2000 males and 1 in 4000 females. FRAXA's goal is to accelerate research aimed at the treatment and cure of fragile X, by direct funding of promising research projects and by raising awareness of this disease.

Report from Washington:

by Mary Beth and David Busby

The Congress

As some of you know, both the Senate and House Appropriations Committees issued their annual Reports to accompany their appropriations to the National Institutes of Health, the Centers for Disease Prevention and Control, and other parts of the Department of Health and Human Services. We are delighted that these Reports, in effect, implemented the fragile X provisions of the Children's Health Act of 2000. This is a real victory for all you FRAGILE X ADVOCATES who wrote and called your Members of Congress! Please write them again now to thank them!

Both Reports encourage the National Institute of Child Health and Human Development (NICHD) to enhance its research efforts on fragile X. The Senate goes a step further by urging the NICHD to provide sufficient funds for "at least three fragile X research centers," and it specifically commends NICHD for teaming up with FRAXA to jointly fund new research.

The House Report also urges the National Institute of Neurological Disorders and Stroke (NINDS) to enhance its research activities on fragile X.

Both Reports encourage the National Institute of Mental Health to support fragile X research in concert with the NICHD and NINDS.

The Senate Report urges the Director of the National Institutes of Health (NIH) to use the Act's new "Pediatric Research Loan Repayment Program to . . . encourage promising investigators to enter various areas of pediatric research, particularly in the areas of Duchenne muscular dystrophy and fragile X."

And, finally, the Senate Report supports the Center for Disease Control in "further research and demonstration projects to facilitate the translation of new scientific knowledge into applied newborn public health screening programs, particularly in the areas of fragile X Syndrome and Cystic Fibrosis."



The National Institutes of Health

On September 7, Katie Clapp and Mary Beth and David Busby met with the Acting Director of the NIH, Dr. Kirschstein, her Deputy, Dr. Maddox, and the Director of the National Institute of Child Health and Human Development, Dr. Alexander. We summarize a pleasant, productive and constructive meeting, as follows:

Dr. Alexander and Dr. Maddox feel that the fragile X research centers can be established most expeditiously and economically as affiliates of presently existing NICHD Mental Retardation and Developmental Disability Centers. Dr. Alexander said that three or more centers would each be funded at up to \$750,000 "direct costs". He announced the following timetable:

- November: NICHD issues Requests for Applications for grants for fragile X Centers.
- March/April: Grant applications are received for review.
- September/October: Successful grant applications are funded.

Dr. Maddox suggested, and it was agreed, that the NIH will host a meeting of fragile X researchers on the NIH campus.

Dr. Kirschstein discussed the implementation of the "Pediatric Research Loan Repayment Program" in the Children's Health Act of 2000. She announced that the Office of Management and Budget has approved program guidelines and that the President's FY 2002 Budget requests money to fund loan repayments for 250 researchers, for the combined Pediatric and Clinical Research Loan Repayment Programs. She expects the funding to increase in fiscal year 2003.

Katie Clapp discussed the state of fragile X research and its relationship with autism research. She reviewed exciting new work being funded by FRAXA and expressed FRAXA's appreciation for the projects being jointly funded by FRAXA with the NIH.

Washington Gala and Lobby Day

Mark your calendars for the fifth annual Mary Higgins Clark Gala, to be held Monday evening, April 29, 2002, at the Four Seasons Hotel in Washington. Co-Chairs: Kitty deChiara, Diane Rehm, and Mary Beth Busby. Host: Roger Mudd. Honoree: Mary Higgins Clark. Dancing to the music of Sydney. Y'all come!

On Tuesday, April 30, FRAGILE X ADVOCATES (that means you!) will have breakfast at the Capitol and then branch out over the Hill to talk to their Members of Congress about fragile X. As those of you who attended the Lobby Day in April of 1999 will remember, that was where our lobby effort got started. We've come a long way!

Here are your Members of Congress who serve on the Health Subcommittees of the Senate and House Committees on Appropriations. They are the key players in funding the fragile X Centers and Researcher Loan program.

Subcommittee on Labor, Health and Human Services and Education Members:

Senate:

Tom Harkin, Chairman,
Iowa
Ernest Hollings, South
Carolina
Daniel Inouye, Hawaii
Harry Reid, Nevada
Herb Kohl, Wisconsin
Patty Murray, Washington
Mary Landrieu, Louisiana
Robert C. Byrd, West
Virginia
Arlen Specter, Ranking
Member, Pennsylvania
Thad Cochran, Mississippi
Judd Gregg, New
Hampshire
Larry Craig, Idaho
Kay Bailey Hutchison, Texas
Ted Stevens, Alaska
Mike DeWine, Ohio.

House:

Ralph Regula, Chairman,
Ohio
David R. Obey, Wisconsin
C.W. Bill Young, Florida
Steny H. Hoyer, Maryland
Ernest J. Istook, Jr.,
Oklahoma
Nancy Pelosi, California
Dan Miller, Florida
Nita M. Lowey, New York
Roger F. Wicker, Mississippi
Rosa DeLauro, Connecticut
Anne Northup, Kentucky
Jesse L. Jackson, Jr., Illinois
Randy "Duke" Cunningham,
California
Patrick J. Kennedy, RI
Kay Granger, Texas
John E. Peterson, PA
Don Sherwood, Pennsylvania

If you live in the state or congressional district of any of the above Members of Congress, please write and thank them for funding fragile X research! Your letters made all the difference in the past and will in the future! Also, make an appointment to visit with them when they are home this Fall. They want to see you (and your vote)!

Fragile X Heroes



Our four staunch champions who sponsored the fragile X Breakthrough Act of 1999, carried its provisions into the Children's Health Act of 2000, and went to bat for us this year before the Senate and House Appropriations Committees were: (clockwise from top left) Senators John Edwards of North Carolina and Chuck Hagel of Nebraska, and Representatives William Delahunt of Massachusetts and Wes Watkins of Oklahoma.

Update on Fragile X Research Centers

On November 9th, Dr. Duane Alexander, Director of NICHD, called to report the following adjustments in the plans for implementing the fragile X Research Centers:

The timetable has changed: Request for applications for Centers will be published by NICHD in December.

The eligibility requirements for submitting an application for Center funding have been clarified: Dr. Alexander reports that only the Principal Investigators of the fourteen Mental Retardation Research Centers (MRRC) currently funded by NICHD will be eligible to apply as Principal Investigators of the new fragile X Research Centers. However, by establishing a collaboration with one of the existing MRRC centers, any investigator at any qualified institution can apply to found and direct a fragile X research center.

r e s e a r c h

“The problem of fragile X is intriguing, because the loss of a single protein causes a variety of behavioral and physical changes,” says Jennifer Darnell, Ph.D. Previously, it was known that the fragile X protein, FMRP, binds to messenger RNA (mRNA) molecules — which carry genetic information (DNA) from a cell’s nucleus to its protein-making machinery — yet the specific mRNAs involved as well as the overall purpose of this protein remained elusive. Now, the researchers present evidence that FMRP may turn up or down the production of certain brain proteins by binding to their mRNA molecules. This type of protein regulation is a crucial aspect of every cell’s life, and in the case of brain cells, is essential for learning and memory formation. The Darnells have identified thirteen mRNAs that FMRP binds, and show that these mRNAs are misregulated in the cells of fragile X patients.

“We found FMRP binding sites in a population of mRNAs shown to be abnormally regulated in fragile X patients,” says Jennifer Darnell. “The proteins coded for by these mRNAs are likely to underlie the problems these patients have.”

Jennifer Darnell identified the mRNA targets by first discovering that FMRP recognizes and tightly binds loop-like structures in RNA, called G-quartets, which represent novel human RNA-binding sites. This finding is intriguing because these structures, which resemble in appearance loose knots along a string, are typically found in DNA and not RNA. After searching a computer database of known mRNAs for the G-quartets, she hit upon a significant finding: many of the mRNAs targeted by FMRP, and their corresponding proteins, play a role in learning and memory, the development of the bones of the face and in the formation of the nervous system — all brain activities involved in fragile X syndrome. In fact, almost all of the thirteen mRNAs identified have biologic functions which fit well with symptoms of fragile X syndrome:

- Six of the mRNAs are associated with the functioning of synapses – the points of contact between brain cells, where information is exchanged between the axon of one neuron to the dendrite of a second neuron. These mRNAs are thought to play roles in maturing and maintaining synapses; at least one is directly linked with learning and memory and another is implicated in the regulation of social behaviors and aggression.

- Three of the mRNAs encode proteins that are involved in growth of neurons: MAP1B is highly expressed in developing neurons, and appears to play an important role in the extension of axons and dendrites. Semaphorin 3F has effects on growth cones and is essential for axon pathfinding. ID3 is expressed in the proliferative zone of the hippocampus that gives rise to granule cells and dentate precursor cells.

- Two mRNAs encode proteins found particularly in brain and testes tissues. One of these, MINT, affects craniofacial development, which may explain why many people with fragile X have a long face and prominent brow. An additional target RNA may be linked with epilepsy; seizures affect some children who have fragile X.

WHAT ARE mRNAs?

mRNAs are the templates that cells use to transform genetic codes (genes) into proteins. From each gene, mRNA is made, and from mRNA, protein is made. The fragile X gene, FMRI, normally produces the protein, FMRP, but in fragile X syndrome, a mutation in this gene results in a lack of FMRP protein.

“It is possible that FMRP is responsible for shuttling certain proteins out to the individual

dendritic spines of neurons, and/or subsequently activating them at the appropriate time during development, as well as during adult memory formation,” says Jennifer Darnell. “This would explain how specific neuronal connections are strengthened to form memories.”

Meanwhile, Steven Warren’s group at Emory also had independently identified mRNA targets of FMRP, using a different technique called microarray, or “DNA chip,” analysis. Robert and Jennifer Darnell met Dr. Warren at the 2001 fragile X Banbury Meeting, the second of a series of annual fragile X research meetings established by FRAXA and funded by the National Institutes of Health and FRAXA. The Darnells and Dr. Warren began collaborating and discovered that nearly 70 percent of Warren’s targets contained the G-quartets.

Using DNA microarray “chip” technology, Warren’s group identified 432 mRNAs from cells in the mouse brain that normally are associated with FMRP. When they compared these to cells derived from people with fragile X syndrome, they identified 251 of those same

update :

mRNAs that were not correctly regulated in the absence of FMRP.

Finally, the researchers demonstrated that the thirteen newly characterized FMRP targets — identified in a test tube in Jennifer Darnell's case — are in fact misregulated in patients' cells, thereby linking their

molecular findings to what's really happening in people's bodies.

Because FMRP plays a role in both the developing and the adult brain, it may eventually be possible to treat some of the symptoms of fragile X syndrome. In addition, the discovery of specific mRNAs involved in the disease has opened the door to new drug targets; it one day may be possible to manipulate the individual

mRNAs or proteins responsible for the symptoms of fragile X, as a means to treat the disease.

For more information, see Rockefeller University press release at www.rockefeller.edu/pubinfo/ment11160/nr.html; original articles in the November 16th issue of Cell (Vol. 107, No. 4).

Research Report: Parent Preferences about Fragile X Screening

*By Don Bailey, Debra Skinner, and Karen Sparkman
Frank Porter Graham Child Development Center
University of North Carolina at Chapel Hill*

Identifying children with fragile X syndrome is a challenging experience for both parents and professionals. As a result, there has been recent discussion about whether systematic screening for fragile X syndrome would be a good policy decision.

Screening for FXS could occur at several points in time. Preconception carrier screening could be offered to women before pregnancy to determine if a woman is a carrier of FXS. [Carrier testing could also be offered to fathers.] Pregnancy carrier screening could be offered to women during pregnancy to determine carrier status. Prenatal screening could determine before birth if a baby has FXS. Newborn screening could identify FXS shortly after birth. And development-based screening could be offered to determine whether a child who is experiencing any developmental or behavioral problems has FXS.

Each of these procedures comes with costs and benefits. Deciding whether to offer screening will be a complicated decision that will include discussions of cost, treatment possibilities, and ethics.

Important to these discussions is how families feel about these options. To determine parent perceptions, we recently conducted a survey of parents of children with FXS to get reactions to different screening options. In collaboration with FRAXA, we mailed written surveys to more than 500 families of children with FXS. The survey asked questions about how



families found out about FXS, the impact of the diagnosis on the family, and opinions about various forms of genetic testing. Questions were developed with input from parents, professionals, and representatives from the Centers for Disease Control. The study was sponsored by a grant from the Office of Special Education Programs, U.S. Department of Education.

We received a great response, with 460 surveys returned that represented 287 mothers and 172 fathers from 299 different families. We are still analyzing the data. Once this process is complete, findings will be submitted for journal publication and posted on the FRAXA web site as well as the Carolina fragile X Project web site. Completed analyses show the following noteworthy findings:

- Only 2% of respondents knew that they or their spouse was a carrier of FXS before getting pregnant
- For all families, the average age of diagnosis of FXS was

57 months. This figure declines significantly for families of children born after 1992, but still remains over 36 months

- More than half of the families had another child before they found out about FXS in their first child
- When asked when is the best time to do genetic screening, 76% said that preconception carrier screening would be their first choice
- Most parents felt that a prenatal or newborn FXS diagnosis would not have a negative effect on parent-child bonding
- More than 90% of families said that if genetic testing showed that their baby was a carrier but not affected, they would still want to be informed.
- More than half said that the diagnosis of FXS affected their decision to have more children

A few parents (4%) felt that screening should never be offered and some concerns were expressed about the consequences of screening. However, most families felt that screening programs would result in a range of positive outcomes for families. We are currently reading and coding the many written comments provided by parents and will summarize this information in forthcoming reports.

This is the first survey of parents to determine their opinions about various forms of genetic screening for FXS. Parents strongly endorsed carrier screening, arguing that this information is needed in order to make informed reproductive decisions. Hopefully this information will be useful as policymakers consider screening options. Because the issue is so complicated, it is unlikely that any form of routine screening will be universally offered in the near future. Thus we will need to keep working with physicians and other professionals to teach them about FXS and encourage testing of children as early as possible.

We appreciate very much the support of FRAXA in conducting this study. The high rate of survey return is rare in survey research. Obviously this means that parents feel strongly about the issues and want their opinions to be heard. Thanks so very much to all parents who participated. We appreciate the time you took to provide information about your lives and your feelings, and we will do our best to make sure that this information is widely circulated to researchers, practitioners, and policy makers.

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defect may somehow protect against cancer, Dr. Soren Schultz-Pedersen of Viborg Hospital, Denmark, and a multicenter team report.

The investigators examined the incidence of cancer in 223 people with fragile X syndrome using the Danish Cytogenetic Registry and the Danish Cancer Registry. Overall, they identified four cases of cancer among the people studied, while almost 11 cases would have been expected based on cancer rates in the general population. The researchers calculated that people with fragile X had only 28% of the cancer risk seen in the general population. Schultz-Pedersen and colleagues point out that in an earlier study of mortality in people with fragile X, only 13 of 83 patients died of cancer, a significantly lower rate than in the general population. "The identification of persons with a decreased risk of cancer opens up possibilities to investigate genetic mechanisms that protect against malignant transformation," the researchers conclude. "Further studies are needed to understand the mechanisms of the ability of the cells to protect themselves against cancer."

This study opens the possibility that some cancer researchers will focus their attention on fragile X, bringing greater attention to our cause. In the meantime, this is good news for families of individuals with fragile X!

FRAXA Booth at Society for Neuroscience Annual Meeting

For the third year in a row, FRAXA sponsored an informational booth at the annual meeting of the Society for Neuroscience. For five days in November, 25,000 neuroscientists converged on San Diego's convention center, including many of the scientists currently supported by FRAXA. This meeting allows us to connect with researchers we know and currently support, and also to introduce fragile X to experts in related fields. Scientists snapped up over 200 FRAXA CDs, a comprehensive multimedia guide to fragile X, including a complete set of FRAXA newsletters, a movie, and texts on education and medication.

This year, Mary Beth and David Busby traveled from Washington, DC to staff FRAXA's booth, and local hero Cindy de Gruchy organized a terrific group of San Diego parents and friends who helped. Thank you to Katie Harris, Hope Busby Burleigh, Shelly Wilson, Carrie Murtagh, Vicky Mulvey, Denise and Jonathan Alvinito, and David Gibson!

Your donation will move research forward

New research project proposals arrived on FRAXA's doorstep on December 1st, and we need your help to fund the best of these! Although the research is heating up, it has been a disappointing year for fundraising; since September 11th, all charities have found it difficult to raise funds for causes unrelated to the tragedy. Contributions to FRAXA over the next two months will determine how many of these exciting new fragile X research proposals can be funded.

Washington Urged to Support Child Health Research

We were pleased that Reuter's News Service reported the following update to news outlets everywhere:

WASHINGTON, Jul 26 (Reuters Health) - Congress needs to immediately renew the law encouraging drug companies to test their products on children, and the Bush administration should fully implement a law passed last year aimed at increasing research on diseases affecting children, a coalition urged at a Capitol Hill news conference Thursday. The Coalition for Children's Health 2001 includes 14 organizations, including United Cerebral Palsy, the Arthritis Foundation and the FRAXA Research Foundation.

"Pediatric research has traditionally been an underfunded medical field," said David Busby of the FRAXA Research Foundation, which supports studies of the genetic disorder fragile X Syndrome. "It is critical that the federal government become more proactive supporting research and encouraging the private sector to take a greater interest in this area of medical research."

The coalition's top priority is passage of the "Best Pharmaceuticals for Children Act," which would reauthorize the law that provides drugmakers with an additional 6 months of market exclusivity for a drug if they conduct clinical trials on children. Without further action, the law expires at the end of this year.

The coalition's other priority is implementation of the Pediatric Research Initiative included in the 2000 Children's Health Act.



Jack and Jacob Massey of Scottsbluff, Nebraska, both have fragile X, but that hasn't slowed them down! This summer, Jack learned to waterski and Jacob won a prize for horsemanship!



Did you know . . .

- You can view detailed financial information about all charities at the website www.guidestar.org. Take a look and you will see that FRAXA's overhead expenses are a mere 6%. We know of no other charity that can top that! If you don't have web access, give us a call.
- This coming year, FRAXA will be part of the Combined Federal Campaign and a number of state workplace campaigns.
- If you have friends and family who might be interested in FRAXA's activities, let us know and we will happily send them our newsletter.

Study Participants Needed in Wisconsin

We are looking for individuals in the 11 to 35 year-old age range with fragile X syndrome to participate in a study researching learning and literacy. Your child should use spoken language as the primary means of communication and know basic shapes and colors. If you are willing to travel to Madison, Wisconsin, you will receive prizes for your child and free hotel accommodations. Contact Mina C. Johnson-Glenberg, Research Scientist at the University of Wisconsin - Madison, email johnsonglen@waisman.wisc.edu., phone: 608/ 262-6768, fax: 608/ 265-4103.

Mina Johnson received FRAXA startup funds for this project last year and has recently received federal funding to continue her work. Congratulations, Mina!

Moms and Kids Needed for Kansas Study

We are researchers at the University of Kansas in Lawrence, KS, studying how young children with fragile X communicate their needs. We are looking for children with fragile X who are 2-6 years old and their mothers to participate in the pilot study for a research project for which we want to write a proposal. We really really need them before the end of the year 2001 as the proposal will be due in February. A family's participation takes about 2-3 hours and can be divided into two sessions. We are offering incentives to families because we know this is a busy time. We reimburse the families for mileage up to \$35 and will give them \$100 in cash at the completion of their participation. Families can come to our site in Lawrence or the Kansas University Medical Center in Kansas City, KS.

If you have children who would qualify or know a family, please contact us or them. We would be glad to answer any questions you may have.

Nancy Brady and Tammy Steeples, Schiefelbusch Institute for Life Span Studies, Wakarusa Research Facility, 1315 Wakarusa Drive, Lawrence, KS 66049, 785-312-5364 or toll free 866-591-3084

Female Carriers of Fragile X Wanted for a Research Study on the Menstrual Cycle.

The Reproductive Endocrine Unit at the Massachusetts General Hospital seeks female carriers of the fragile X premutation for a research study to examine the menstrual cycle. Mothers of children affected with fragile X and any other women who are fragile X carriers are invited to participate. The study will help determine whether there are changes in the menstrual cycle hormones in women who carry the fragile X premutation. The study also involves the option of participating in neurological and psychological testing to examine thinking and personality traits. Women should be age 18-50 yrs. Up to \$50 stipend. Call Patty at 617-726-5387.

About Tissue Donation

Human tissue donated at the time of surgery or death by people of all ages, or in the case of miscarriage or pregnancy termination, is a precious resource on which researchers depend. FRAXA and the Brain and Tissue Bank for Developmental Disorders in Maryland have produced a joint brochure about fragile X tissue donation. If you would like a supply of these brochures for your support group meeting or family members, please call Doreen DiMeglio, (800) 847-1539, at the bank, or Katie Clapp, (978) 462-1866 at FRAXA.

Fragile X Listserv, in Spanish and Portuguese

This forum is for sharing personal and professional experiences and opinions about fragile X Syndrome. Over 140 professionals and family members from all the Spanish and Portuguese-speaking countries are currently subscribed. All spanish/portuguese-speaking persons from any country in the world are welcome to join.

To subscribe, send mail to: xfragil-subscribe@onelist.com

To unsubscribe, send mail to:
xfragil-unsubscribe@onelist.com

To send messages, send mail to: xfragil@onelist.com

To reach list owner, send mail to:
xfragil-owner@onelist.com

URL of this page:
<http://www.onelist.com/community/xfragil>

FRAXA Gets a New Volunteer

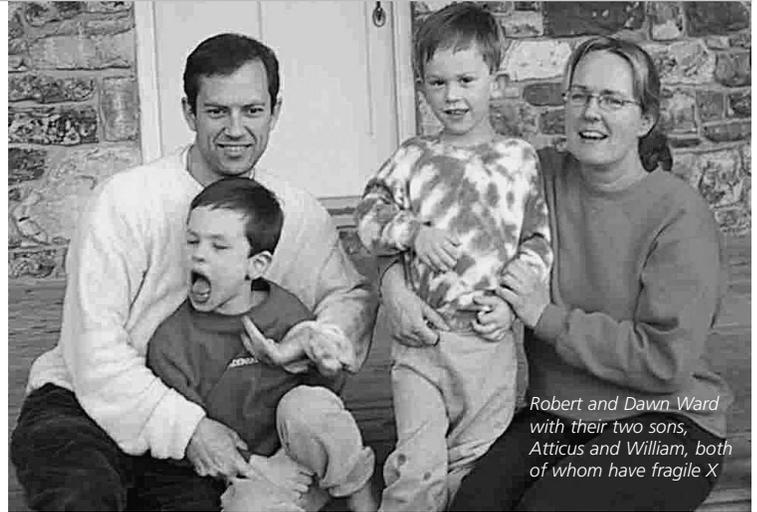
Hi! My name is Dawn Ward and I am the parent of two boys, ages 5 and 7, who have fragile X. Now that they are in school full-time this year, I have begun working as a volunteer for FRAXA.

When my children were at home during the day, I didn't have enough energy or time to really even think about volunteering; I was just coping with our family's very stressful day-to-day life. But now that my days are "free", it is with great satisfaction that I sit down to my desk and computer in my newly created home office (I moved my boys into the same bedroom). There's nothing else I'd rather be doing. I have every hope and belief that a cure will be found for fragile X and I intend to speed up progress toward that day!

You can help FRAXA by letting me know of any ideas that you have, but just don't have the time or energy to pursue alone. My email address is CureFragileX@aol.com and my phone number is 703-631-1845. I am presently working on:

- organizing an annual Walk for Fragile X at various sites around the globe;
- putting together a fundraising guide to help those who want to fundraise but need some help on how to;
- and anything that Katie Clapp sends my way.

I look forward to hearing from you!



Robert and Dawn Ward with their two sons, Atticus and William, both of whom have fragile X

Would you like to start a support group or organize an event?

As FRAXA's mailing list has grown, chances are that we might know of families who live nearby but don't know each other. If you would like us to help you connect with others in your area, send an email, call, or drop us a line. Let us know whether you are a parent (grandparent, friend, etc.) and if we have permission to share your name with others in your area.

Update from the National Fragile X Foundation

I'm pleased to announce the first titles in our "special topics pamphlet series," Females and Fragile X and Fragile X and Sexuality, are now available. Both are the result of collaboration between NFXF staff and advisors. A third pamphlet, Behavior Management and Fragile X, will be available in the near future. All three deal with subjects that are related to common questions that the NFXF receives. Like all NFXF pamphlets, these are designed to be an introduction to a specific topic. They are intended to help parents and professionals formulate questions relevant to their specific concerns, and as a starting point for further learning. Each includes references to resources that address the topics in greater detail. Additional titles will be released in the months to come.

I'm also pleased to announce an exciting new endeavor called the Education Project. This project is designed to help teachers better include children with fragile X within the regular classroom. The project is a collaborative effort

of the NFXF, parents from the NY and NJ support groups, Dr. Vicki Sudhalter, Dr. Marcia Braden and others.

The final product will be produced in a loose-leaf binder and will address preschool through young adulthood. It will include sections on:

What is Fragile X?

General information about fragile X, characteristics, and learning styles.

Adapting Curriculum

Recommendations, suggestions and guidelines regarding adapting curriculum and lesson plans.

Examples of Lesson Plans

Gathered from parents and teachers across the country, and at the July, 2001 Chicago Conference.

Please contact me with any questions or comments.

Robert Miller, Executive Director 800-688-8765

NATLFX@FragileX.org www.FragileX.org

FRAXA EVENTS

5th Annual Fragile X Golf Benefit

The 5th Annual fragile X Golf Benefit was held on Monday, July 30, 2001 at the Shaker Heights Country Club in Shaker Heights, Ohio. The event, with AT&T as the Title Sponsor, was a major success raising over \$100,000 again!

The 156 golfers enjoyed the challenging golf course and were then joined by an additional 150 guests after the tournament. The three hundred attendees enjoyed appetizers, drinks and the large variety of exciting Silent Auction items.

During the dinner program, Dr. Michael Tranfaglia of FRAXA Research Foundation spoke of the rapid progress being made in fragile X research. He also recognized the importance of Dr. Alan Tartakoff's research at Case Western Reserve University here in Cleveland. This grant is funded primarily from the proceeds of this benefit. We showed a video entitled "Hope for the Future" which told the audience about fragile X and the promising research projects underway.

Special guest, Doug Dieken (former Cleveland Browns player & current radio announcer) and Honorary Chairpersons, Herb & Nancy Score conducted a fun and entertaining Live Auction.



Ara Bagdasarian, Jay Bagdasarian, and Larry Karobiwian man the golf leader board

Our core committee of Leslie and Ara Bagdasarian, Jeanne & Mike Sydenstricker, Rod Tyler and Jim Vitalie were joined this year by more volunteers from the fragile X Alliance of Ohio, friends and family members. Special thanks go to Kristie Braley and Conferon, a local meeting planning company, who provided support and volunteers to help with this event.

A fun day was had by all, but we cannot lose sight of the reason behind this benefit – to raise awareness and research funds for fragile X Syndrome – the most common inherited form of mental impairment and learning disabilities worldwide. If you would like a copy of our program or have any questions, please email Leslie at lbagdas@oh.verio.com.

Nascar Racing and the Civitans support FRAXA

Dear FRAXA,

I am a member of the local Civitan Club and have a 4-year-old son with fragile X. I recently gave a talk to the club about fragile X and they generously gave me a check for your foundation. Developmental delays and mental retardation are the primary interests of the Civitan Clubs nationwide, so it is very appropriate that the money be given to this cause. This photo is of me accepting the check from the President of the Richmond Civitans, Inell Allen.



A few of the many volunteers

This money was raised by selling concessions twice a year at the North Carolina Motor Speedway in Rockingham, NC. These popular Nascar races usually attract 30,000 people per day, so it is hard work, but a lot of fun!

Thank you from me, the Richmond Civitans, and from my son, David, for all of your hard work. Hopefully we can continue our support in the future.

– Sarah Tamura
Rockingham, NC



Stone Pony Party in Asbury Park

In October, New Jersey couple Denise and John Sabo hosted a benefit

for FRAXA Research Foundation at The Stone Pony. The Sabos have a 3-year old son Kyle, who has fragile X. There was music by "The Soul Engines," stand-up comedy by "Dr. Sensitivity" Joe Picolli and Otto and George, and a performance by Elvis impersonator Angel Pastrana. FRAXA friends came from as far away as Virginia (thank you, Carol and Brian!) and Massachusetts to celebrate the event.

Pennies from Heaven!

Jen Nardo of Hockessin, Delaware, persuaded officials at a local mall to donate coins dropped in their fountain to FRAXA. Jen reports:

I just received the coins from my area mall. I was able to get one of the five crates of coins counted and I am going to try to wrap the rest with some help. This could add up to almost \$1000 for FRAXA! I just wanted to let everyone know that there indeed are "pennies from heaven!"

Jen is organizing a fundraiser for February 9th at a local church. There will be entertainment, hors d'oeuvres, cash bar, and a silent auction; anyone who would like to join in can call Jen at (302) 234-7854 or email jen9612@aol.com.

Upcoming Gala in New England

Springtime is celebration time for FRAXA! Join us for an evening at the Corinthian Yacht Club, on the seashore in Marblehead, Massachusetts, on Thursday, May 16th. Plan a long weekend and explore the history and beauty of Marblehead in the spring. This event takes place two weeks after our Washington, DC, Mary Higgins Clark Annual Gala, which is on Monday, April 29th – come join us for both!

If you can help recruit sponsors, both corporate and individual, or if you would like to reserve a table for the evening, please contact Leslie Eddy at (781) 631-9196 or Katie Clapp at FRAXA.

Available from FRAXA:

All prices include shipping within the U.S. Please call or e-mail for international orders.

FRAXA CD

One CD-Rom holds FRAXA's video Unlocking Fragile X, publications on educational strategies and medications, newsletter issues, fragile X articles and FRAXA's brochure (Acrobat format). Works with PC or Macintosh computers. Upon request with any donation.

FRAXA "X" Lapel Pin

Gold-plated FRAXA logo pin is a wonderful gift! \$10

FRAXA Umbrellas

Pop-up umbrellas in an assortment of colors, with FRAXA's logo in white. This is a terrific gift for teachers and friends. \$12

Unlocking Fragile X

An emotional, inspiring look at fragile X, FRAXA and current research, with author and grandmother Mary Higgins Clark, Nobel Prize Winners James D. Watson, Ph.D, and Eric Kandel, MD, and many others. This 10-minute video is a great fundraising aid. \$8

FRAXA Tribute and Memorial Cards

Both are available in packages of 10 cards for \$30.

FRAXA T-Shirts

White, all-cotton T-shirts feature FRAXA's logo on left chest. Adult sizes: M, L, XL, XXL. \$12

Fragile X: A to Z

Edited by Wendy Dillworth, this is chock full of stories from daily life with fragile X children. Browse through helpful suggestions on topics such as adolescence, bike riding, and dental work. 73 pages, \$15.

A Medication Guide for Fragile X

By Michael Tranfaglia, MD, Psychiatrist and Parent. This guide helps parents and others understand behavioral symptoms of fragile X and the medications commonly prescribed to help manage these symptoms. \$20.

Educating Boys with Fragile X

By Gail Spiridigliozzi, Ph.D., this guide has specific helpful suggestions aimed particularly at teachers and therapists. 20 pages, \$10.

Free: FRAXA Brochures and Gift Envelopes

Fragile X Information Cards

Many families have asked for a card that they can give to people who have no knowledge of fragile X. Business-size cards: \$10 per 100.

FRAXA POSTDOCTORAL FELLOWSHIPS REQUEST FOR GRANT APPLICATIONS

**Upcoming Deadlines:
May 1, 2002 and December 1, 2002**

FRAXA offers fellowships and grants to encourage research aimed at finding a specific treatment and ultimate cure for fragile X syndrome:

- Postdoctoral fellowships of up to \$35,000 each per year
- Investigator-initiated grants for innovative pilot studies aimed at developing and characterizing new therapeutic approaches (no funding limit)

FRAXA is particularly interested in preclinical studies of potential pharmacological and genetic treatments for fragile X and studies aimed at understanding the function of the FMR1 gene. Applications are accepted twice each year. Information is available at www.fraxa.org or by contacting FRAXA.

Calendar of Events

MONDAY APRIL 29

5th Annual Mary Higgins Clark Fragile X Gala, at The Four Seasons Hotel, Washington, DC. Chaired by Diane Rehm, Kitty de Chiara, and Mary Beth Busby. Call Mary Beth at 202-462-2323 to reserve tables or for sponsorship information.

TUESDAY APRIL 30

Fragile X Lobby Day in Washington, DC. The morning after the gala, we will fan out across Capitol Hill to meet with Members of Congress. Please contact Mary Beth Busby at 202-462-2323

THURSDAY MAY 16TH

Black Tie Gala, at the Corinthian Yacht Club, Marblehead, Massachusetts, with celebrity guests. Help us fill the club! Chaired by Leslie Eddy; call her at 781-631-9196 to join in.

Additional events are planned in New York, OHIO, Massachusetts, and Maryland – stay tuned!

FRAXA
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FRAXA UPDATE

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FRAXA would like to thank Networx of Newburyport, MA for hosting the FRAXA website and email. Networx has donated this important resource for the past 6 years

PLEASE HELP
FRAXA
in supporting research aimed
at treatment for fragile X RESEARCH
FOUNDATION

FRAXA is a national 501(c)(3) tax-exempt organization. Every penny you donate goes to research: FRAXA has specific grants to cover all overhead. Supporters receive this newsletter and are welcome to participate as active volunteers.

Yes, I would like to help FRAXA

- | | |
|---|---|
| <input type="checkbox"/> Member (\$25+) | <input type="checkbox"/> Benefactor (\$500+) |
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| <input type="checkbox"/> Sponsor (\$100+) | <input type="checkbox"/> Named Research Fund (\$5000+) |
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