X Marks

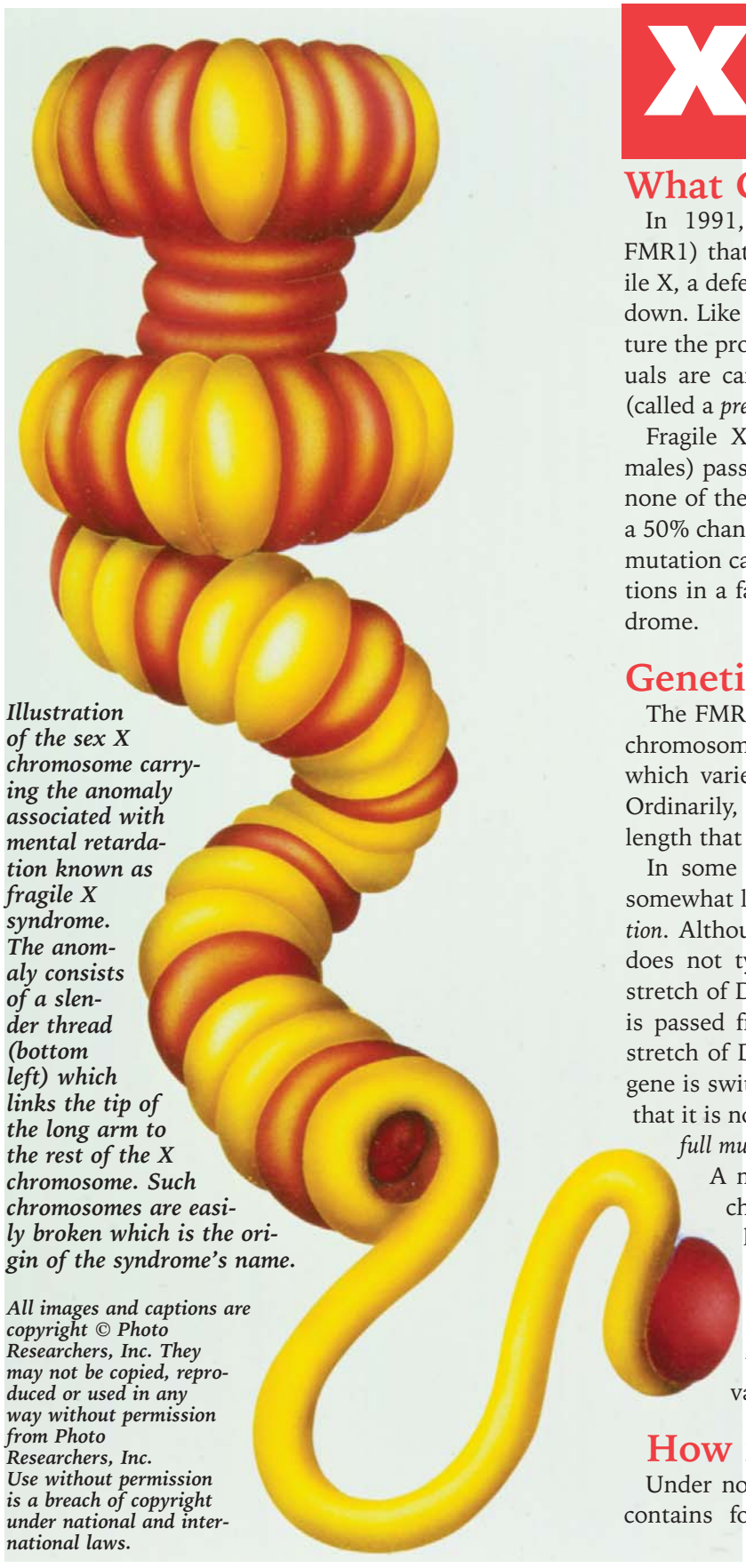

Illustration of the sex X chromosome carrying the anomaly associated with mental retardation known as fragile X syndrome. The anomaly consists of a slender thread (bottom left) which links the tip of the long arm to the rest of the X chromosome. Such chromosomes are easily broken which is the origin of the syndrome's name.

What Causes Fragile X?

In 1991, scientists discovered the gene (called FMR1) that causes fragile X. In individuals with fragile X, a defect in FMR1 (a *full mutation*) shuts the gene down. Like a defective factory, FMR1 cannot manufacture the protein that it normally makes. Other individuals are carriers: they have a small defect in FMR1 (called a *premutation*) but do not show symptoms.

Fragile X is inherited. Carrier men (transmitting males) pass the premutation to all their daughters but none of their sons. Each child of a carrier woman has a 50% chance of inheriting the gene. The fragile X premutation can be passed silently down through generations in a family before a child is affected by the syndrome.

Genetic Cause of Fragile X

The FMR1 gene is located on the long arm of the X chromosome. Within this gene lies a region of DNA which varies in length from one person to another. Ordinarily, this stretch of DNA falls within a range of length that would be considered *normal*.

In some people, however, this stretch of DNA is somewhat longer; this gene change is called a *premutation*. Although a person who carries the premutation does not typically have symptoms of fragile X, the stretch of DNA is prone to further expansion when it is passed from a woman to her children. When the stretch of DNA expands beyond a certain length, the gene is switched off and does not produce the protein that it is normally makes. This gene change is called a *full mutation*.

A male who inherits a full mutation exhibits characteristics of fragile X syndrome because his only X chromosome contains the mutated gene. A female may not be as severely affected as a male because each cell of her body needs to use only one of its two X chromosomes and randomly inactivates the other.

How is Fragile X inherited?

Under normal circumstances, each cell in the body contains forty-six (twenty-three pairs of) chromo-

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the Spot

somes. These chromosomes consist of genetic material (DNA) necessary for the production of proteins which lead to growth, development and physical/intellectual characteristics. The first twenty-two pairs of chromosomes are the same in males and females. The remaining two chromosomes are called the sex chromosomes (X and Y). The sex chromosomes determine whether a person is male or female.

Males have only one X chromosome which is inherited from the mother at conception. They receive a Y chromosome from the father. Females inherit two X chromosomes, one from each parent.

What Are the Common Symptoms of Fragile X?

Symptoms of fragile X include: mental impairment, ranging from learning disabilities to mental retardation, attention deficit and hyperactivity, anxiety and unstable mood, autistic-like behaviors; long face, large ears, flat feet, hyperextensible joints, especially fingers. Seizures (epilepsy) affect about 25% of people with fragile X.

Boys are typically more severely affected than girls. While most boys have mental retardation, only one-third to one-half of girls have significant intellectual impairment; the rest have either normal IQ or learning disabilities. Emotional and behavioral problems are common in both sexes.

About 20% of boys with fragile X meet full criteria for autism. Most boys and some girls have some symptoms of autism, but many tend to be very social and interested in other people.

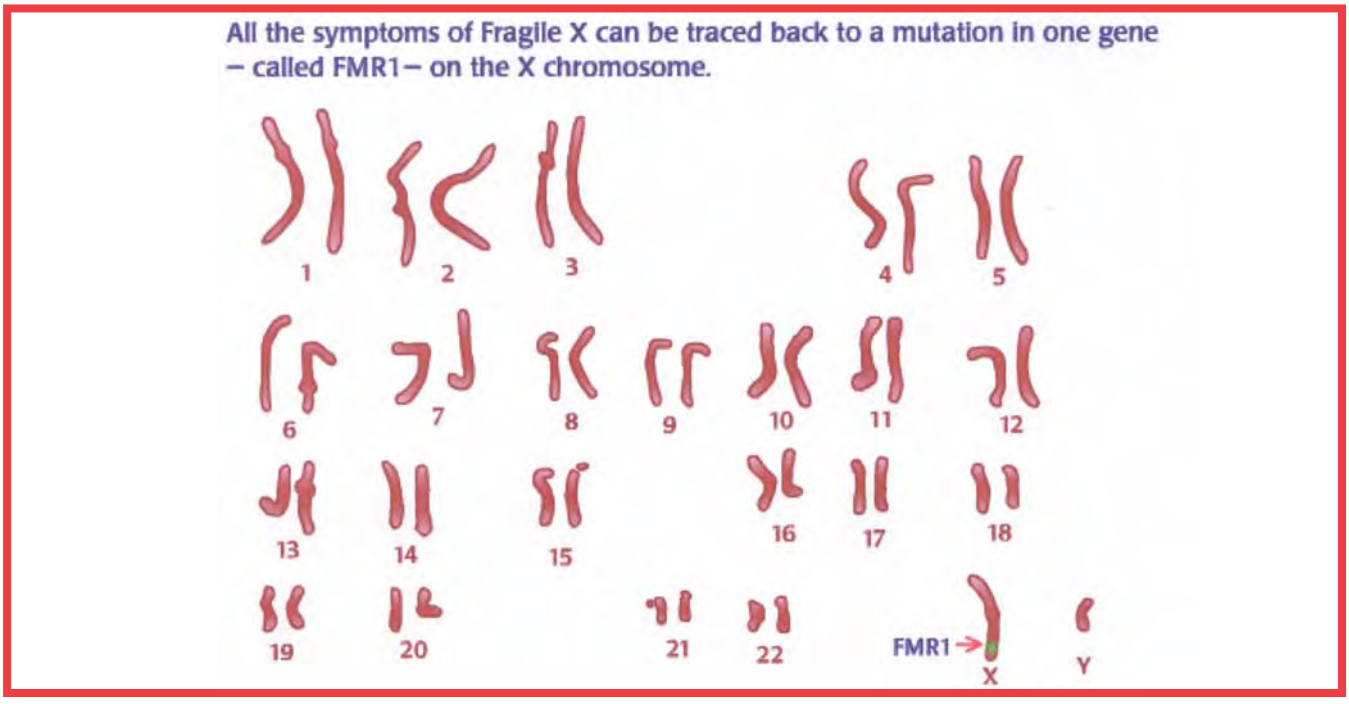
Is There a Test for Fragile X?

A DNA based test to diagnose fragile X was developed in 1992. This test is quite accurate, and it can detect both carriers and fully-affected individuals. The blood test that can be ordered by any physician. The blood sample is then sent to one of the laboratories that offers the test. It usually takes several weeks to get the results.

Because the symptoms of fragile X can be quite subtle, especially in young children, and because it is so frequent in the general population, many medical specialists recommend that testing for fragile X be considered for any individual with otherwise unexplained developmental delay or mental retardation.

Most major medical centers in the United States now offer the DNA test for fragile X. This test costs about $200 but may be covered by health insurance. For more information about testing, talk to your doctor or genetic counsellor.

Information and chart (this page) reprinted with permission from the Fragile X Research Foundation website at www.fraxa.org.



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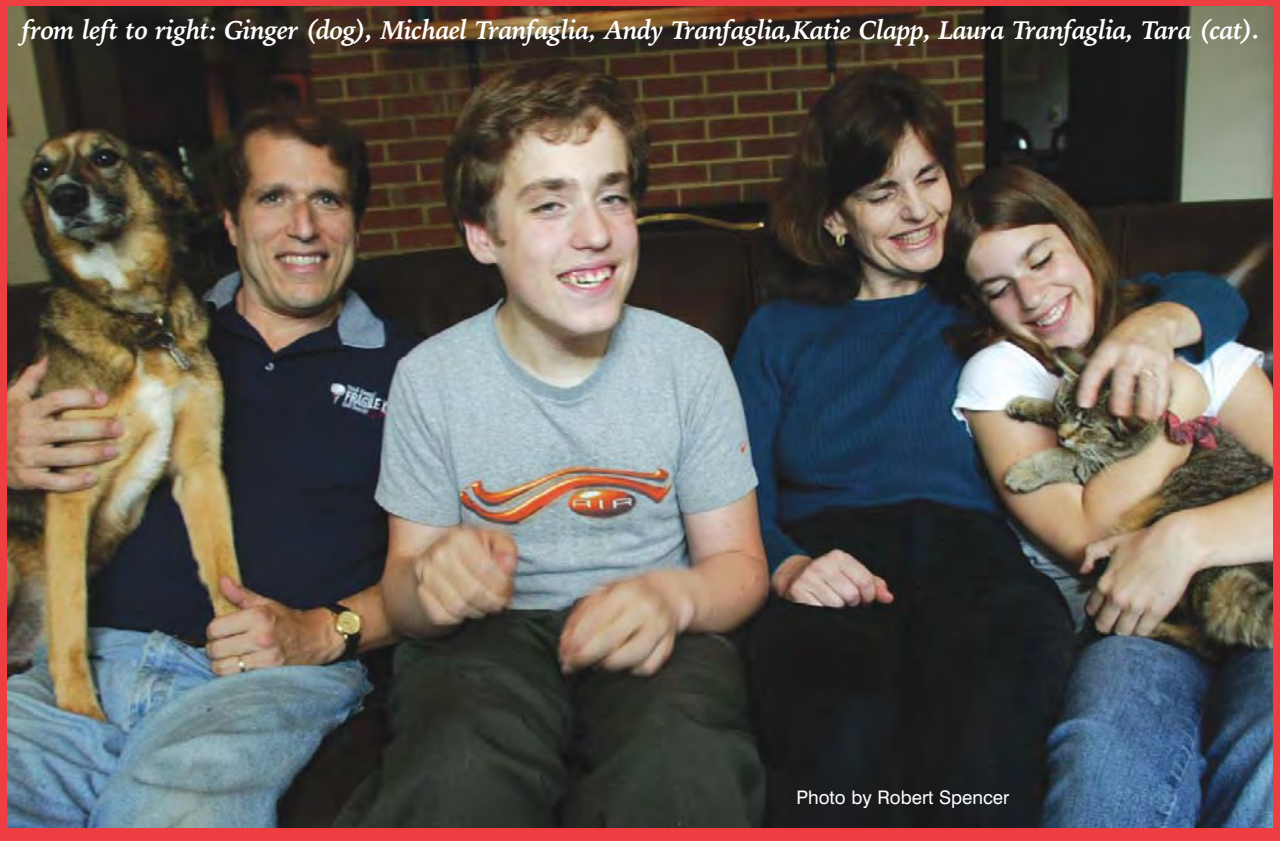
A Different Life

Conversation with Founder of The FRAXA Research Foundation



from left to right: Ginger (dog), Michael Tranfaglia, Andy Tranfaglia,Katie Clapp, Laura Tranfaglia, Tara (cat).

Photo by Robert Spencer

The search for answers, for a treatment, for a cure to fragile X has been "an absolute mission for me," says Katie Clapp President and co-founder of the FRAXA Research Foundation. She established the not-for-profit organization in 1994 with her husband, psychopharmacologist Michael Tranfaglia. The two are parents of Andy, 15, and Laura, 13, both of whom have fragile X syndrome.

Headquartered in Newburyport, MS, the organization, today raises more than $2 million a year, most of which is awarded in the form of grants to university-based researchers. "We are not looking for the perfect cure," says Ms. Clapp. "Our mission is to fund research that will result in treatments for this generation and for everybody else in the future."

For Ms. Clapp and Dr. Tranfaglia, the drive to find answers is personal. "We had problems with Andy from day one," says Ms. Clapp. "He had lots of feeding problems and he didn't do much of anything. He didn't crawl until he was 21 months old. But we were first time parents, and we didn't know for sure. Half the time the medical professionals would say he was fine. By the time Andy was a year old, however, he had received a diagnosis of devel-

opmental delays. When Ms. Clapp became pregnant with her second child, "I remember saying, 'Please look carefully and tell me if I need to be worried about genetics.' The doctors said 'no'." Neither Andy nor Laura, received a diagnosis of fragile X until after the family had moved from South Carolina to MA. "The first time we walked into the developmental pediatrician's office with Andy, the doctor knew it was fragile X. Andy had the look—the ears and the flapping hands. He was tested that day." Several weeks later test results confirmed fragile X. "That's when we had our daughter tested," says Ms. Clapp.

Reminiscent of the movie, "Lorenzo's Oil," Katie Clapp and Dr. Tranfaglia wrote countless letters to try to interest bio-technology and pharmeceutical companies into taking on the challenge of finding a cure for fragile X syndrome.

The couple immediately dove headlong into research. "We started calling around for information. We learned that fragile X is the most common cause of inherited mental impairment. Two weeks after the diagnosis, we went to Canada to attend a conference on fragile X, and that is where we got really immersed in the whole thing. We realized it is just one gene that is shut down and the consequence is that there is a protein missing. We started thinking, 'Well, can we reintroduce the protein?'" At that point, Ms. Clapp says she learned that just a handful of researchers were probing for solutions. "That's when it hit me—not enough research was being done." At about the same time, "My husband went off to the medical school library and started reading. He would come back excited about prospects for gene therapy. So that's when we decided to try to do something about it."

Reminiscent of the movie, *Lorenzo's Oil*, Katie Clapp and Dr. Tranfaglia wrote countless letters to try to interest bio technology and pharmeceutical companies into taking on the challenge of finding a cure for fragile X syndrome. "We tried for almost a year, but we didn't succeed in getting a company to take it on as a profitable enterprise." Undaunted, the two persevered for another four years while Ms. Clapp continued teaching computer science and working as a computer consultant. Ultimately, though, she found that she could not handle both her career and her mission. "I hit a kind of crisis," she says. "In three weeks time, I got three traffic tickets. I knew I needed to stop. I quit the computer stuff."

Dr. Tranfaglia's story is somewhat similar. As a psychopharmacologist, he worked on the hospital staff in Newburyport, joined a group practice and later became a consulting psychiatrist in nursing homes. In the remaining hours he tried to help grow the research Foundation. Finally, in the summer of 2004, he decreased the number of hours spent as a consultant and took on the nearly full time positions of Medical Director and Treasurer of FRAXA—positions he had been handling in an unofficial capacity since 1994.

The couple's dedication to the cause has yielded results. FRAXA boasts 4,000 members, a 10 member Board of Directors comprised of parents of children with fragile X, a Board of approximately 30 scientific advisors and an 11 member Parent Advisory Board. Twice yearly, the organization accepts research proposals and awards grants to university based researchers.

"As parents of a child with fragile X, you do have to be proactive. You have to learn a lot first so that you will know. It is enlightened self interest."
–Katie Clapp

Ms. Clapp notes that researchers are exploring fragile X from various angles. While gene therapy remains a bright, long-range possibility, FRAXA has lately been investing more heavily in basic research, "because the basic research is uncovering

what is wrong, and that makes sense to me as a mother," she says. "I see my child, I see what he does and how he responds. Drug companies are working on treatments that will correct what is wrong." As scientists discover the pathways within the brain, psychiatric drugs used to treat problems become more specific. A child with fragile X syndrome, may, for instance suffer from seizures. Medication to handle seizures enables the child to improve his or her ability to focus, which in turn improves the ability to learn. "More and more, we are finding that research into fragile X provides a link to what scientists believe is going on with other disorders such as Prader-Willi syndrome, autism, Retts syndrome. We are finding evidence that there are common pathways. I am exited about finding treatments for people with fragile X syndrome, but I will also be very excited if we can help uncover treatments for other disorders."

> ***"Our secret is that my husband and I have found something we could do—an avenue for our various emotions."***
> ***–Katie Clapp***

Despite the countless hours and the inevitable frustrations of heading up a research foundation, Ms. Clapp is grateful to be involved. "Our secret is that my husband and I have found something we could do—an avenue for our various emotions. As parents of a child with fragile X, you do have to be proactive. You have to learn a lot first so that you will know. It is enlightened self interest," she states. "As long as you are doing something, you are not going to get cynical, or exhausted or waylaid along the way. It kind of becomes your center. There is not anything harder than being a special needs parent. But is funny how, because you have to do it—because there is no way out—being the parent of a special needs' child doesn't make for a miserable life. It makes for a different life."

FRAXA Research Foundation, 45 Pleasant Street, Newburyport, MA 01950. ❄

Unraveling the Mystery

Author Mary Jane Clark discusses life as the parent of a child with fragile X syndrome

"Writing was total therapy for me," says author Mary Jane Clark. "I felt like so many things in my life were out of control. In this fictional world I made up, I could control everything that happened. How the people looked, what they said. I could make things turn out all right at the end—or not—but I decided. It was cathartic."

Mary Jane Clark

Shortly before she began writing novels, Ms. Clark's son, David, now age 16, was diagnosed with fragile X syndrome. He was seven months old. "Actually, when I brought him in for has three-month check up, the pediatrician told me his eyes weren't following the way they should have been." Although he used the term low muscle tone, "I think the pediatrician suspected something even more. He said, 'If he hasn't progressed when you bring him in next month, I think you should take him for some testing.' The next month, when I came in, the pediatrician already had the name of three places for us to go. One of those places was Hackensack University Medical Center." Several weeks later, a geneticist called and asked that Ms. Clark and her (then) husband come in for a visit. "We sat there just kind of stunned as they explained what they had found."

David began receiving therapeutic services immediately. At the age of three, his parents enrolled him in a pre-school. Today, he is 6' 2" tall, "incredibly strong and healthy as can be," says his mother. Although he is quite verbal, David cannot read or write, and "He has a lot of anxiety." Despite the deficits, "He is very intelligent," says his Mother. "He has an incredible memory for faces and names. If you came here one day and talked to him for ten minutes and came by again six months from now or a year or two years later, he would remember your name. Unfortunately, though, what has happened to David happens to too many children with disabilities. He has a lot of solitary time. He spends a lot of time watching television and movies. He enjoys it, and I let him do it, but I wish he had a fuller life with more activities that he enjoyed."

While writing novels has become the centerpiece of

her career, Ms. Clark continues to work part time as a writer/ producer at CBS. "I still enjoy television, and I have a history at CBS. I started working there as a desk assistant right after college." Then, too, Ms. Clark notes, "Writing novels is very solitary. It is hard work and there are lots of times when I'd rather be doing anything else but writing. I love *having written*. I love it when it is done. It not unlike pregnancy," she explains. "Pregnancy is tough, but once the baby is born, it's very satisfying."

Ms. Clark is the former daughter-in-law of the popular author Mary Higgins Clark. The two maintain "a cordial, family relationship, mostly through the children," says the younger Ms. Clark. "We keep our writing separate."

Without a doubt, having David in her life has brought Ms. Clark enormous joy. But she is realistic. "You cannot short sell the difficulties of rearing a child with a disability. Sadness and disappointment "come to you in the weirdest ways," she says. "Babyhood was very, very tough. Then I started to think, 'O.K. I've got a little handle on this.' But there was that phase when David was ten or twelve years old, and I'd drive on a highway and see boys his age playing soccer, or I'd be at someone's wedding and see the young groom dancing with his mother. … The sadness," she says, "does not necessarily come about at graduation or a birthday party—it happens out of nowhere—suddenly there will be a flash of what it could have been and what it is. I love David, and I would not trade him for anything in the world. But I do wish he had a different life. Absolutely."

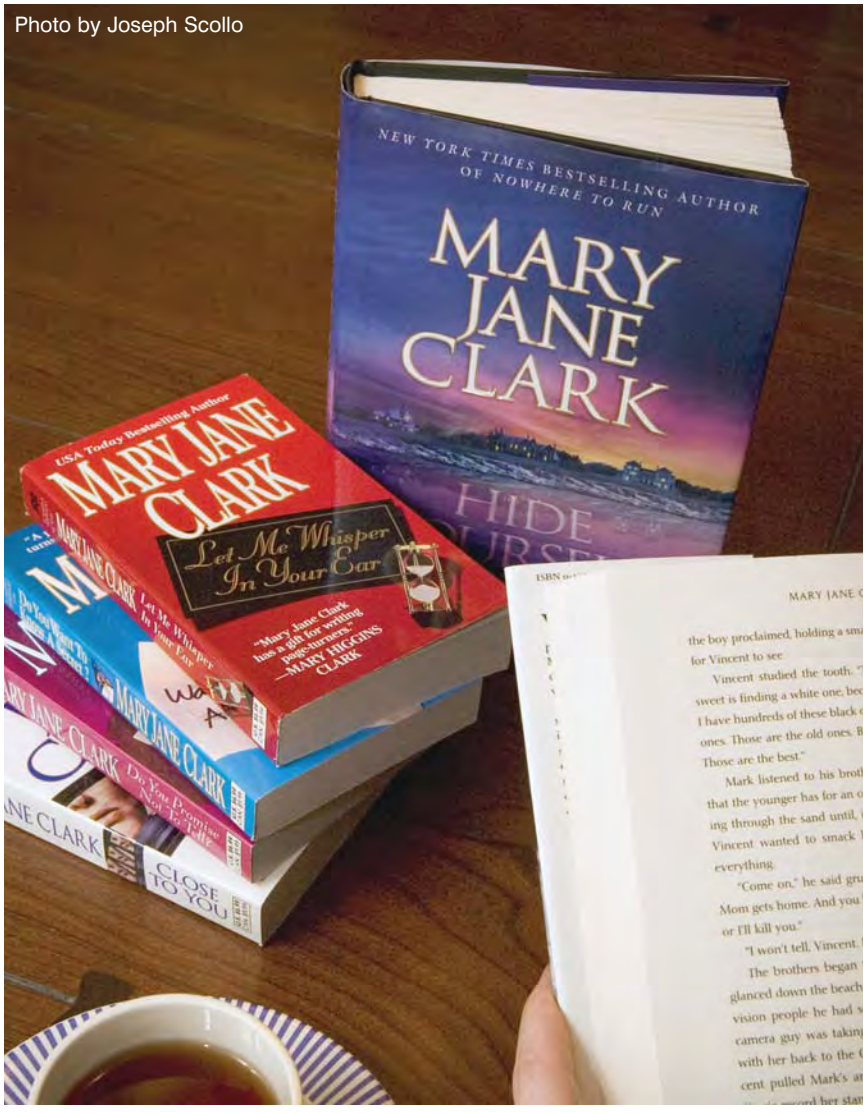


Photo by Joseph Scollo

> ***"Having a child with a disability makes me want to write about that. I feel as though the novels can be good educational tools."***
> ***–Mary Jane Clark***

According to Ms. Clark, David's having fragile X has affected the whole family. Daughter Elizabeth, 20, "has a depth at this young age that I don't think she would have had otherwise. Elizabeth is not a carrier of fragile X" she is quick to add. "So she won't be dealing with it, thank God." Ms. Clark says too that both she and Elizabeth's father "have tried very hard to make sure that Elizabeth does not get swallowed up by fragile X—that she gets the attention and individual time she deserves—that her whole life is not about David."

Although the Clarks divorced when their children were quite young, Ms. Clark says, "I do not blame fragile X and David's situation for my marriage falling apart." However, she adds, "The diagnosis is pretty much like an earthquake. It is something seismic that

shakes up your life and only the buildings with the strongest foundations are left standing."

The author of seven books, all published by St. Martin's Press, Ms. Clark recalls the early years when she would write at odd hours over long periods of time. "I started thirteen years ago. The first book took two years for me to write, two years to rewrite, and it sat on a shelf for two years. Because the kids were little, I was writing when they went to bed at night and early in the morning or when they were with their father. I would write page by page whenever I could get it done. Now I write one book a year."

> ***"I love David, and I would not trade him for anything in the word. But I do wish he had a different life. Absolutely."***
> ***–Mary Jane Clark***

Savvy fans will know that several of Ms. Clark's books include characters with disabilities. Her first book, entitled *Do You Want to Know a Secret?* incorporates a child with fragile X syndrome who holds the solution to the mystery. Another novel features a child with cystic fibrosis. "I am sensitive to anyone with a disability or chronic condition," she says. "Having a child with a disability makes me want to write about that. I feel as though the novels can be good educational tools."

As she looks ahead to her son's future, Ms. Clark considers the possibility of a community residence. "If we can find the right place, I think a community residence is a good idea. As the child gets older, it gives him or her a little independence. I would like to feel that David can make his way in the world. But my biggest fear is what will happen when I am gone."

Ms. Clark is affiliated with the FRAXA Research Foundation. She has appeared several times on network television discussing fragile X syndrome. Each of her last four books includes a dedication that reads: "And again, for Elizabeth and David. And for all those who struggle with fragile X syndrome, the most common inherited form of mental impairment. Please God. Let us find a treatment or cure."

❄

Facing F

Photo by Joseph Scollo

Janet and Michael Armetta with Paul, left, and Jacob

ragile X

The Armetta Family

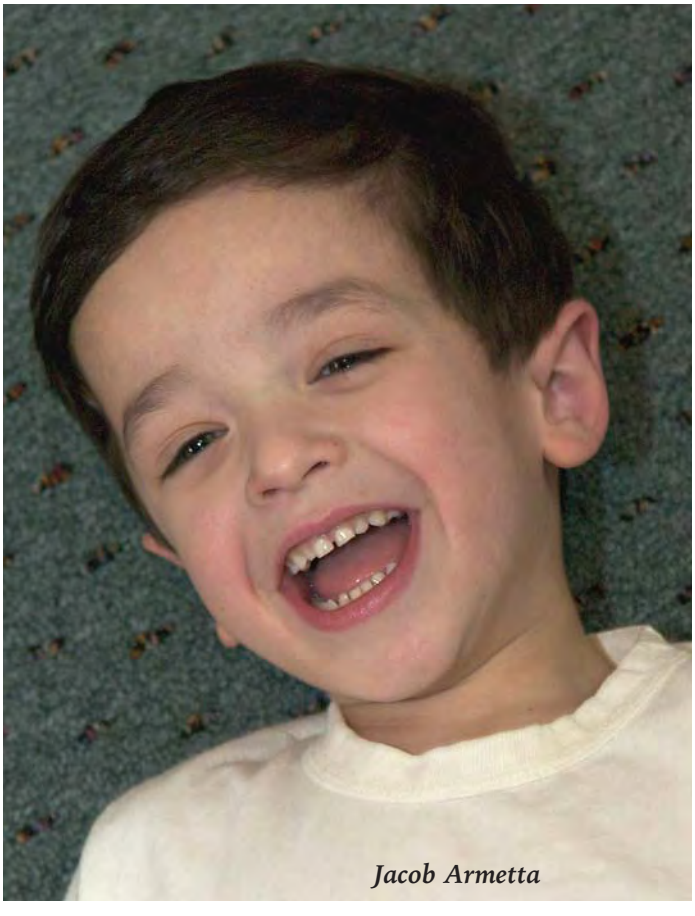
Janet Armetta offers a word of advice to doctors: "When speaking to the mothers and fathers of children with newly diagnosed or as yet undiagnosed disabilities, be gentle with us. We are the fragile ones."

Ms. Armetta and husband Michael are the parents of five-year-old Jacob and two-year-old Paul. At the age of three, Jacob was diagnosed with fragile X syndrome. As is often the case with first time parents, Ms. Armetta said that some early signs of delay went unnoticed. He was a "slow sucker," she recalls. He didn't hold his head up too early. When he wasn't sitting up by nine months, the Armettas voiced their concerns to the pediatrician. Somewhat reluctantly, the pediatrician agreed that Jacob could see a pediatric neurologist. "We saw the neurologist when Jacob was ten months old." The neurologist did not seem unduly concerned. Still within the parameters of normal development, Jacob began sitting several weeks later.

"The neurologist turned to us and said, 'Do you think he has autism?' Other than Rain Man or afternoon TV specials on disabilities, what the heck did I know?" –Janet Armetta

Sporadically during the next several years, the Armettas experienced vague concerns. "If we called his name, he didn't pay attention, he barely flinched. My family kept insisting it was his hearing. When he would hurt himself, he rarely cried. I almost want to say he had a higher tolerance for pain then most people. It was almost like his nerve endings didn't kick in for a while…like he wasn't getting sensory input from his brain."

Although he crawled at a typical age, he did not walk until 22 months. Mr. Armetta thought perhaps Jacob's wearing shoes was inhibiting his ability to walk. "The doctor said shoes or no shoes wouldn't make a difference," recalls Ms. Armetta.



Jacob Armetta

"When he was 15 months old, though, I was more concerned with his lack of speech than his lack of walking. Yet the neurologist took a *wait and see* stance and suggested that Jacob return for a visit at 18 months. It was the 18-month visit that prompted Ms. Armetta to advise doctors to go gently. "The neurologist turned to us and said, 'Do you think he has autism?' Other than *Rain Man* or afternoon TV specials on disabilities, what the heck did I know?" asks Ms. Armetta. Still without a diagnosis, Jacob began receiving Early Intervention services at home. By age two and ½ the Armettas enrolled their son part time at Prime Time for Kids Early Learning Center. Finally, when Jacob was three, a blood test confirmed the diagnosis of fragile X syndrome.

Today, Jacob has a "huge vocabulary," notes his mother. Although his speech is not always clear, "I can understand most of what he says. Cognitively, he understands a lot and physically, he is very healthy. I don't know if you can outgrow low muscle tone, but Jake has incredible balance." As is the case with every child, Jake "has his meltdowns," says his mom. "Maybe the meltdowns are more extreme than they would be for a child who doesn't have fragile X." On the whole, Jacob Armetta is doing well. "If he had to have something wrong with him, I'm glad that it is something that keeps him smiling and happy most of the time. He is a favorite among the teachers. He's a favorite among people."

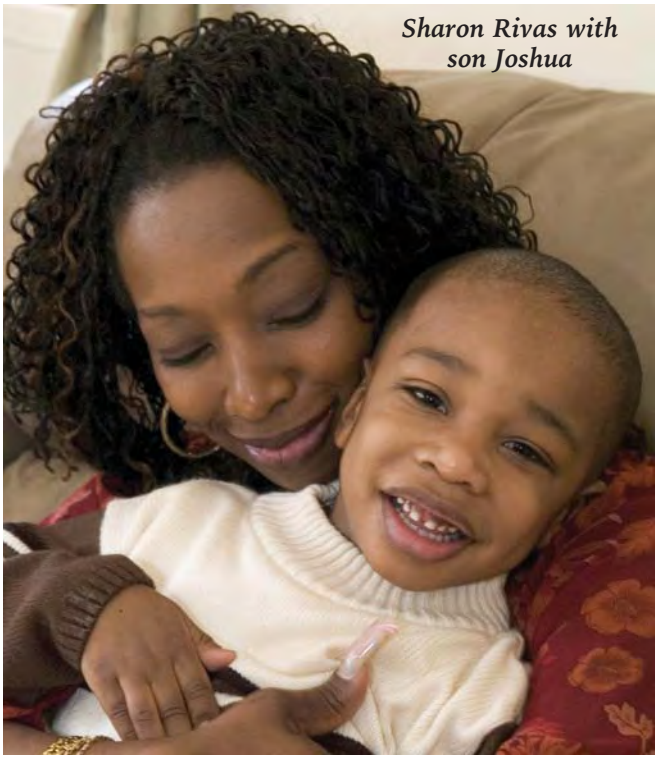
Ms. Armetta has high expectations for Jacob. "We have stated a higher education fund for Jacob. If he can go to college he will, if not, then he will do something else. One day I would like him to be able to live on his own and support himself. He is friendly, happy and sociable. He has a good sense of humor. People just warm up to him. It has always been this way."

The Armettas live in Chestnut Ridge, New York. Mr. Armetta is a retired officer with the New York Police Department. Ms. Armetta recently gave up her job in the garment center so that she could spend more time at home with her children.

While she hopes that science will find a cure for fragile X, Ms. Armetta does not count on that. Instead she takes one day at a time. "I am the glass half full type of person. There are times when it gets frustrating, but I am glad that Jake is doing well. I guess he makes things easy on us."

The Rivas Family

When Joshua Rivas was not making any intelligible sounds by the age of one and one half, the pediatrician suggested that Sharon and Nelson Rivas take their son to a neurologist for a full behavioral evaluation. The neurologist ordered a blood test to check for fragile X syndrome.



Sharon Rivas with son Joshua

"I had never heard of fragile X until Joshua was diagnosed with it," recalls Ms. Rivas. "Anytime someone is diagnosed with a serious condition you get kind of worried, and Joshua was young." Rather than allow fear to overwhelm her, almost immediately, Ms. Rivas began to gather as much information as possible about fragile X syndrome. She quickly learned that severity of the condition varies with each child. She learned too, that children with fragile X syndrome often exhibit autistic like tendencies.

At the time of Joshua's diagnosis, the Rivas family was living in New Jersey. Shortly thereafter, Joshua started receiving physical, occupational and

speech therapies at home through Early Intervention services. Because fragile X syndrome is a genetic disorder, the neurologist suggested that Joshua's sister, Kaetlin, just a year younger than her brother, be tested for fragile X. That test too proved to be positive. Following her diagnosis, Kaetlin began receiving speech and occupational therapies at home through Early Intervention services. In 2003, when the family moved to Nyack, New York, both Joshua and Kaetlin were enrolled in Prime Time for Kids.

Although neither Joshua, now four years old, nor his sister, three, is verbal, their mother says, "They comprehend just about everything. Recently both children underwent a surgical procedure through which tubes were placed in their ears to prevent frequent infections. "One of the doctors told me that the fluid in their ears had been making it sound as though people were speaking to them from under water." Ms. Rivas says that since the surgery, both children are making a greater effort to speak. "My hope is that eventually they will have speech because at times, communication can be difficult."

Ms. Rivas says she is "extremely happy with Prime Time for Kids." For the moment the Rivas' have not arranged for supplemental therapies beyond those that the children receive during school hours. Both parents work full time. Ms. Rivas works in the technical operations at Novartis Pharmaceuticals in Suffern. Mr. Rivas is a supervisor at Wyeth Pharmaceuticals in Pearl River. "In the future we may look into arranging for additional therapies after school at home," says Ms. Rivas. This mother believes that her own positive attitude goes a long way towards helping her children. "I went through so much to have these kids. And fragile X or not, they are my kids. If I think in a negative way, if I think, 'Why did this happen to me?' or 'Why did this happen to them?', then I won't be any good to them. I have to stay positive. I have to try to do whatever I can do. If you look at my children you would not know that there was anything wrong with them. They walk, they jump, they play. They get into everything. They may not be learning things at the rate that most children are learning them. It may take them longer, but I see that they are learning and progressing. They are doing it in their own special way."

Kaetlin Rivas

The Schindelheim Family

When Michelle Schindelheim received her son Gavi's diagnosis "I was very depressed," she says. "I made millions of phone calls and one led to the next. You learn things as you trip over them. I would speak to as many people as possible. What was important was finding the best occupational therapists, physical therapists, chiropractors, speech therapists, food regimen. I would keep notes, and one person would lead me to the next person."

Eight-year-old Gavi is a student in the school-age Applied Behavior Analysis (ABA) program at Prime Time for Kids Early Learning Center. Gavi's sister Erica is ten years old. Having a brother with fragile X syndrome has "definitely affected Erica," says the children's mother. "Erica is very bright, but by the end

Michelle and Marty Schindelheim with Gavi, and Erica

of first grade, she wasn't reading."

Ms. Schindelheim recommends enrolling the brothers or sisters of children with disabilities in sibling support groups. Difficult as it may be to cope with the added stress, "Parents cannot negate the needs and activities of the other children in the family. I did that for a while. I think most people have a tendency to do that.

"When you have child with a disability, you have to work on your marriage. You may have to go for counseling," says Ms. Schindelheim. Even those parents who are familiar with disabilities may face unexpected challenges. Mr. Schindelheim is a guidance counselor for children with special needs. Ms. Schindelheim teaches English as a second language. "At different times we've each been depressed, and at other times we've thought things looked positive. You have to appreciate your child for who he is, see his strengths and take one day at a time. You have to try to enjoy life," states Ms. Schindelheim.

Creating a relatively calm atmosphere in their Riverdale, New York home requires a concerted effort. "I had to learn to balance and not search for a thousand remedies for fragile X. It hurts everyone. My husband felt slighted. My daughter was affected. After a while you realize that you do your best and what is going to happen is going to happen. You realize that your kid is never going to be like every other kid. He's wonderful. He is just a different kid."

Enduring stares, questions, sometimes even taunts are part and parcel of rearing a child with a disability. Learning to cope with such situations is vital. Nonetheless, the lack of sensitivity on the part of the general public can be disquieting. "Sometimes my husband Marty will look at Gavi and say, 'Poor Gavi.' And I will say to him, 'Not poor Gavi, it is the people who don't see Gavi for what he is that are poor.' For good or for bad, when you are out in public you are going to be noticed. I am too busy now to worry about people staring at us or feeling sorry for us."

As is the case with so many parents of children with disabilities, Ms. Schindelheim has frequently received compliments about the strength of her character—compliments that imply that only capable people are given the privilege of rearing children with disabilities. "I was strong to begin with," she says. "I didn't need this to make me stronger. Everyone has problems. You just can't always see them. No one's life is smooth."

"I had to learn to balance and not search for a thousand remedies for fragile X."
–Michelle Schindelheim

On the power of positive thinking, Ms. Schindelheim is an expert. "Never listen to anything negative," she says, "especially when spoken by neurologists." Too often, she notes, "neurologists tell you every limitation. Someone once told us to put our child in an institution. I never thought Gavi would walk or talk or be potty trained. Now I'm thinking that he will read."

"Most parents of children I know walk away crying after seeing a neurologist. You have to be positive. Intelligence cannot always be measured on traditional tests. Don't listen to the limitations that people place on your child. Fight for your kid, because if you don't fight for your kid, who else will? You are his advocate. If you feel your kid is great, he'll go in that direction."

Gavi Schindelheim

The Muhlmeister Family

"Initially, I was calm," says Jody Muhlmeister. "I am a psychologist and I had read all the books. I knew very early on that there were issues. He had trouble with rolling over, trouble with sitting. I brought it to the doctor's attention when Marc wasn't meeting the milestones. The doctor suggested that he get physical therapy as soon as possible. We wanted to make sure that we addressed the muscle tone. I thought, 'So, he has low muscle tone.' I was told by the physical therapist that he might not be a world-class athlete. That didn't upset me too much. As I started noticing all of the other deficiencies, I became more and more upset and nervous—I wouldn't use the word panic—until I heard the word fragile X. I switched pediatricians when he was a little over a year old."

Ms. Muhlmeister and husband Alex are the parents of three-year old Marc who has fragile X syndrome and Talia, one and ½. The family lives in Congers, New York. Marc is enrolled in the Applied Behavior Analysis (ABA) program for children with autism and related disabilities at Prime Time for Kids Early Learning Center. The ABA program focuses on improving a child's ability to communicate. Although Marc's verbal vocabulary is 'sporadic', he is learning to sign. Sociable and loving, Ms. Muhlmeister says her son is also 'distractible, and he has oral motor issues. He chews on things," she says. Nontheless, Marc's mother continues "Cognitively I feel that he is in tact. He likes to

learn, and that is a very big plus."

Occasionally described, as a 'veil' covering the essence of the child, fragile X, say some parents, appears to hide their offsprings' true abilities. "The future is a big question mark and it scares me," admits Ms. Muhlmeister. "Our greatest hope for him is that he can be independent, but we don't know." At this stage of their son's life, the Muhlmeisters are learning the ropes of advocacy. Beyond the services Mark receives at Prime Time for Kids, the Muhlmeisters have made a 'huge financial commitment' to provide additional therapies at home for their son.

Prior to the age of three, the determination, type and duration of services required by a child with a developmental disability is made by the Department of Health. After the age of three, a committee of qualified individuals within the child's school district decides the type and duration of services required by the youngster. Not infrequently, parents must advocate for services they believe are necessary for their child or children. Some parents hire attorneys to convince the *powers that be* of the need for more services. When all else fails, parents have to "really dig into your pockets," says Ms. Muhlmeister. "And there is never enough."

As they face the inevitable struggles ahead, Ms. Mulmeister finds herself grateful for the advice of professionals at Prime Time For Kids and for the friends she has made there. "Just being able to talk to somebody else who is experiencing this, to be able to connect with someone else is good," she comments. She has also joined the National Fragile X Foundation (see story page 43) where a hotline gives her immediate answers to some of her questions and concerns. On the home front, she relies heavily on her husband. "He has been very good," she says. "In some ways he has been better at dealing with fragile X than I have." A financial analyst by profession, "He is very matter of fact. He is not as emotional as I am. He is really the one who has Marc saying the words he does say. My husband is very persistent, and that has been a huge asset."

To parents who suspect a problem, fragile X or otherwise, Ms. Muhlmeister urges that they not "close their eyes to the signs. Doctors sometimes say the children will 'grow out of it,' to 'wait and give it time.' If you are concerned, though, don't give it time, have the children evaluated. If there is nothing wrong, then you will have wasted an afternoon.

"The hardest part of fragile X is knowing it has been in our family and that we never knew," says Ms. Muhlmeister. "There is some guilt." ❄

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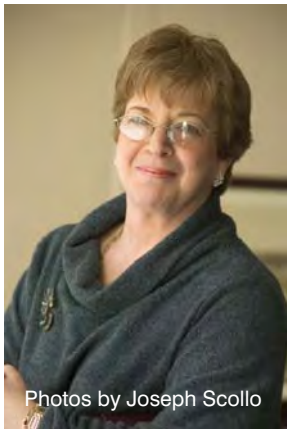
Dr. Vicki Sudhalter, Institute for Basic Research



Photos by Joseph Scollo

The Language Link

Fragile X Research at Staten Island Based Institute

Established through the New York State legislature in 1966, the Institute for Basic Research (IBR) is the research component of the New York State Office of Mental Retardation and Developmental Disabilities (OMRDD). IBR consists of departments and laboratories that conduct basic and clinical research in the field of developmental disabilities and mental retardation. The complex is located adjacent to the campus of the College of Staten Island. In addition to conducting basic research, the Institute also provides clinical services and educational programs for individuals with developmental disabilities.

"The research we are doing here will help people with fragile X syndrome live more content and fulfilled lives, and that is what is important," says Dr. Vicki Sudhalter, a research scientist and Head of the Clinical Psycholinguistics Laboratory at the Institute for Basic Research. "I see my job here as helping to maximize the ability of individuals with developmental disabilities to enjoy what life has to offer—helping them to taste more of life, see more of life, to live more complete lives."

Echoing Dr. Sudhalter's statement is Deborah Sturm Rausch, OMRDD Director of Public Affairs. "The Institute for Basic Research is dedicated to the mission of OMRDD which is to understand mental retardation and developmental disabilities from a scientific point of view in order to prevent and treat all aspects of mental retardation and developmental disabilities."

In her 20 years at the IBR, Dr. Sudhalter has collaborated with experts in numerous disciplines to explore the underlying "behavioral characteristics that make people with fragile X syndrome different from people with other cognitive disabilities," she says. "One of the beauties of the IBR is that there are geneticists and behavioral scientists working together here." Among the findings documented by Dr. Sudhalter and her colleagues is the fact that, while the language difficulties faced by people with fragile X syndrome often appear to be similar to those with autism and other disabilities, the underlying reasons for those difficulties stem from different sources. Therefore, the treatment of language difficulties arising from different disabilities should not necessarily be the same. "Each person is an individual and everyone's needs are different. We cannot lump people together as being delayed, and we are not going to lock step these people into treatments."

"The interdisciplinary process of science at the Institute for Basic Research presents a tremendous advantage," notes Ms. Rausch. "Each individual brings different training and different approaches to be woven into the internal structure of the IBR."

Thirteen years ago, Dr. Sudhalter began to focus her attentions on the psycho physiological aspects of certain developmental disabilities. (Psycho physiological refers to that system that deals with the correlations between the mind, behavior and bodily mechanisms.) We began to think that *sensory integration deficits* and the overreaction to those deficits could be a very important cause for many of the language, cognitive and behavioral problems commonly seen in people with fragile X syndrome," she explains. "It may also be possible that these overreactions cause such individuals to experience high anxiety in the presence of certain environmental stimuli, such as bright lights, loud sounds or crowded spaces. This understanding has proved to be helpful in creating more effective therapies"

Knowing the root cause of a person's language problems has helped Dr. Sudhalter and

others determine ways to help mitigate or overcome problems. Deficits in sensory integration may interfere with one's ability to perform on the job. People who do not have fragile X syndrome will recognize the symptoms of an over-or-under-activated nervous system. Dr. Sudhalter explains, for example, that, "During a typical day at the office, most people will, at one point or another, need to stand up and stretch, drink a cup of coffee or take a walk down the hall to *depressurize*. People with fragile X syndrome need to depressurize on the job too, but they might not know how to make that happen. In such a situation, the agitation they experience will affect their ability to speak or perform appropriately. With training and time, people with fragile X syndrome might ultimately learn to recognize such signs as sweating palms or a rapidly beating heart and ask for what they need to become more comfortable. Or, an astute supervisor will notice such indications as agitation, fidgetiness or red ears and suggest that the person take a break, have some tea or a walk from one side of the office to the other."


Deborah Sturm Rausch, OMRDD

It has been said that some people are more suited to one career than another. A facility with math, for instance, may encourage one to become an accountant. The possession of a beautiful voice might encourage one to think about a career in the opera. For people with sensory integration deficits, some jobs may be more suitable than others.

"For instance, in more rural communities where farming is an option," says Dr. Sudhalter, "people with fragile X syndrome have been known to make great farmers. The need to get sensory input into the soles of the feet is one reason why a person might enjoy tilling fields." The repetitive motion and the physical labor are aspects of the job in which a person with fragile X syndrome can take pleasure. Similarly, young people with fragile X syndrome enjoy fishing or working with horses. The combination of physical labor and quiet are plusses to the person with sensory integration deficits.

When working with young children, Dr. Sudhalter says, "It is essential to educate the educators. So often we look at the child with a developmental disability as having a *behavior problem*. But we need to realize that the problem may actually be that we do not really understand how to help the child. By better understanding the underlying cause of problem behavior in a child with a developmental disability, we are able to more effectively help the child, and he or she will be a happier, more contented person. One of our missions at the Institute for Basic Research is, in fact, to help people who care for people with disabilities. Frequently, parents will come to us because they want us to help them help teachers. A small part of what we do is visit classrooms to see what it is about the classroom setting that is helping or not helping a particular child who has fragile X syndrome. Following the visit, we talk with the teachers and the parents. Our goal is to help each individual reach his or her fullest potential. We don't know what that potential is until we have created the best environment for someone with that particular disability."

Although fragile X syndrome has lately been the topic of magazine and newspaper articles and television talk shows, the experts say the greater press may not be a reflection of a rising incidence of fragile X syndrome. "As we continue to perfect science, we are better at documentation," notes Ms. Rausch. "There is also an increased awareness of fragile X among the lay population and among physicians." Research relative to fragile X syndrome is regularly published in medical journals to which physicians have access.

Whether it is fragile X syndrome or other developmental disabilities, agencies such as ARC of Rockland are "helping the general public to understand that each person is unique and brings to this world something special about him or herself," says Ms. Rausch. "We are continuing to see progress in humanity. We see an increased awareness as community leaders embrace people with disabilities." From the vantage point of OMRDD, "We see possibility and opportunity for all people." *For additional information contact the NY State OMRDD Office of Public Affairs at (518) 474-6601.* ❄

Creating A Lifetime of Abilities For People with Disabilities

ARC LIGHT

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Tribute to Martin Luther King