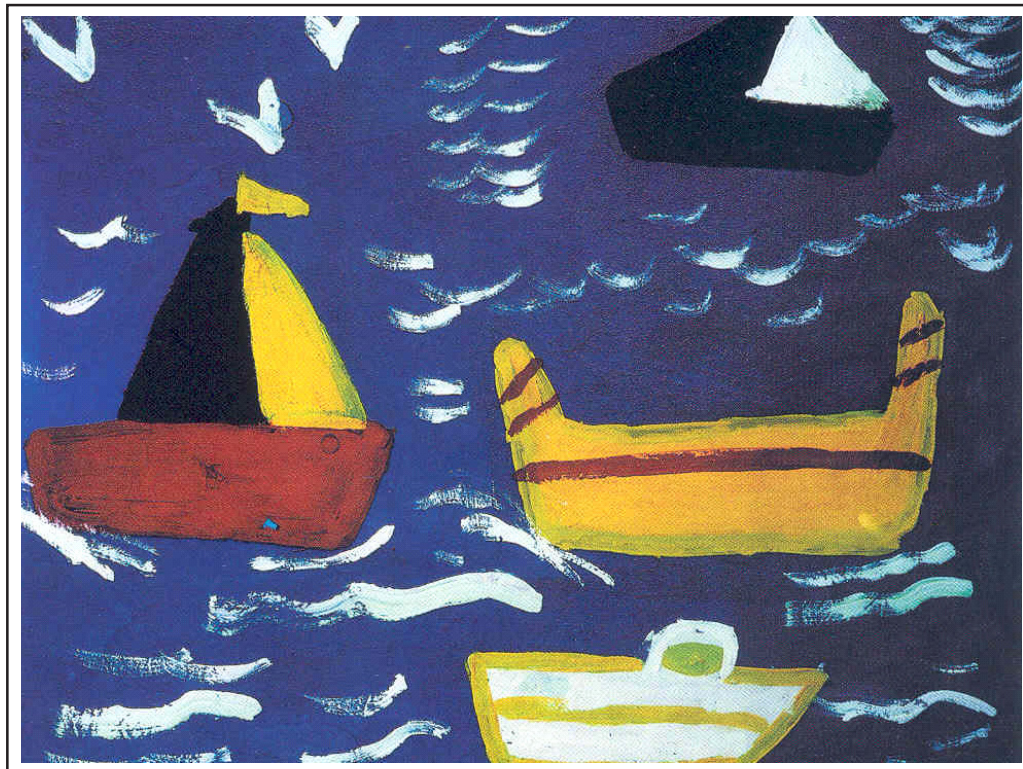


**Waisman Center**  
**University of Wisconsin – Madison**

*Dedicated to the advancement of knowledge about human development, developmental disabilities, and neurodegenerative disease.*

## **Study on Family Adaptation to Fragile X Syndrome**



### **Report 1: Overview of the Study and Shared Experiences from Mothers**

- Principal Investigators -

Marsha Mailick Seltzer, Ph.D.  
Leonard Abbeduto, Ph.D.  
Jan S. Greenberg, Ph.D.

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# Table of Contents

## **Section I: Context for the Study**

Introduction.....	1
About fragile X syndrome.....	1
What is the purpose of this study?.....	2
How did we find the families who are participating in this study?.....	3

## **Section II: Description of the Families in the Study**

Who are the families in our study?.....	4
Who are the adolescents and adults with FXS in our study?.....	6

## **Section III: Comments from Mothers**

Pathways to diagnosis of FXS.....	8
Understanding and coming to terms with FXS.....	10
My son or daughter's strengths.....	12
My son or daughter's challenges.....	14
Impact on siblings without FXS.....	16
Parenting struggles.....	18
Experiences as a mother.....	20
Concerns and plans for the future.....	22

## **Section 1: Context for the Study**

### **Introduction**

We want to thank you and your family for participating in our study “Family Adaptation to Fragile X Syndrome.” Without your participation, we would not be able to conduct this important research. This report is the first in a series of reports that you will be receiving that describe findings from our study.

The first two sections of this report provide an overview of our study and a description of the families who are participating. In the third section, we have assembled some of the comments mothers shared with us during the first round of interviews. These comments highlight the extraordinary struggles and challenges associated with having a son or daughter with fragile X syndrome (FXS). They also show the tremendous rewards and triumphs experienced by these families.

The diverse nature of FXS is reflected in the comments of mothers in this report. In part, this diversity is due to the X-linked pattern of inheritance of FXS. Females with FXS have a milder pattern of impairments than males. Whereas males with FXS generally have moderate to severe intellectual disability, one-third to one-half of women with FXS have average intellectual functioning. Families who have a daughter with FXS may have very different experiences than those who have a son. In addition, there is considerable variability within each gender. Given this diversity, some of the comments in this report may reflect your own family experiences. Other comments may surprise you and highlight the differences between families who have a son or daughter with FXS.

### **About Fragile X Syndrome**

Fragile X syndrome is most common inherited cause of intellectual disability and the only known cause of autism spectrum disorders. It is estimated that FXS occurs in about 1 in 4,000 males and 1 in 8,000 females. Fragile X syndrome results from a mutation in the 5' untranslated region of the Fragile X Mental Retardation (*FMR1*) gene located on the X chromosome. The *FMR1* gene typically has less than 55 repeats of the CGG sequence of nucleotides. Individuals who have 55 to 200 CGG repeats in the gene are said to be carriers of the “premutation.” Individuals who have more than 200 CGG repeats are said to have the full mutation of FXS.

Individuals with FXS generally have cognitive and communication impairments and co-occurring conditions including attention problems, hyperactivity, anxiety, and autistic symptoms. These impairments and co-occurring conditions are often less severe in females with FXS as compared to males. This gender difference is because females have two X chromosomes (only one of which is affected), whereas males only have one. In addition, X inactivation in women can result in a mosaic pattern of affectedness. This means that in women one of the X chromosomes is turned off early in development. In some cells, the X carrying the expanded *FMR1* gene is turned off. In other cells, it's the chromosome carrying the non-expanded (or normal) version of the gene that is turned off.

### **What is the purpose of the study?**

Our 5-year study focuses on families who have an adolescent or adult diagnosed with the full mutation of FXS. There is little research on the experiences of families who have a son or daughter with FXS or the experiences of individuals with FXS during their adolescence or adulthood. The purpose of our study is to learn about the well-being of mothers and their sons or daughters with FXS, the day-to-day stress and care responsibilities experienced by mothers, and the impact of having a son or daughter with FXS on the broader family environment. An additional aim of the study is to learn about the quality of life of adolescents and adults with FXS in regard to their service needs, social and recreational activities, friendships, and family relationships. Families in the study will be interviewed three times (every 18 months) over 5 years. This will allow us to understand the stability and change in the lives of mothers and their son or daughter with FXS over time.

This study is funded by the National Institute of Child Health and Human Development (NICHD) and is part of the national network of NICHD-funded research centers on FXS. The other center sites are located at the University of North Carolina at Chapel Hill and the University of Kansas. Together, we are collaborating on an integrated and longitudinal set of three studies, with each site focused on a different aspect of family adaptation to FXS. Researchers at the University of North Carolina are conducting a 5 year-long study to develop newborn screening procedures for FXS and to understand the ramifications of this screening for families. Researchers at the University of Kansas are conducting a 5 year-long study to understand how mothers interact with their younger child with FXS and to evaluate the effectiveness of an intervention aimed at improving the responsiveness of mothers for the outcomes of the child with FXS. Our study at the Waisman Center, University of Wisconsin-Madison, is unique in that it is the first large-scale

longitudinal study to be focused exclusively on families of adolescents and adults with FXS. We hope that the knowledge we gain from our studies will help improve life for individuals with FXS and their families.

### **How did we find the families who are participating in this study?**

To date, 148 families with an adolescent or adult son or daughter (aged 12 years or older) are participating in the study. In order to qualify for the study, mothers had to live with their son or daughter or have at least weekly contact with them.

Documentation confirming that the son or daughter has FXS was required.

Families were recruited through advertisements to listservs and websites maintained by national organizations supporting families with FXS, brochures distributed at parent and professional conferences and to health professionals, national research registries, and by contacting families participating in other FXS research projects. Interested families were informed about the study over the phone and mailed an informational packet inviting them to participate.

## Section II: Description of the Families in our Study

### Who are the families in our study?

- Mothers range in age from 35 to 79 years, with an average age of 50 years and their husbands range in age from 32 to 72 years, with an average age of 52 years.
- The large majority of mothers and fathers (96% and 97%, respectively) are Caucasian, with the remaining parents being African American and American Indian.
- The number of children per family ranges from 1 to 6, with an average of 2.4 children per family. More than one-third (37%) of the families have multiple children with FXS. Figure 1 below shows the percentage of families in the study by number of children with FXS.

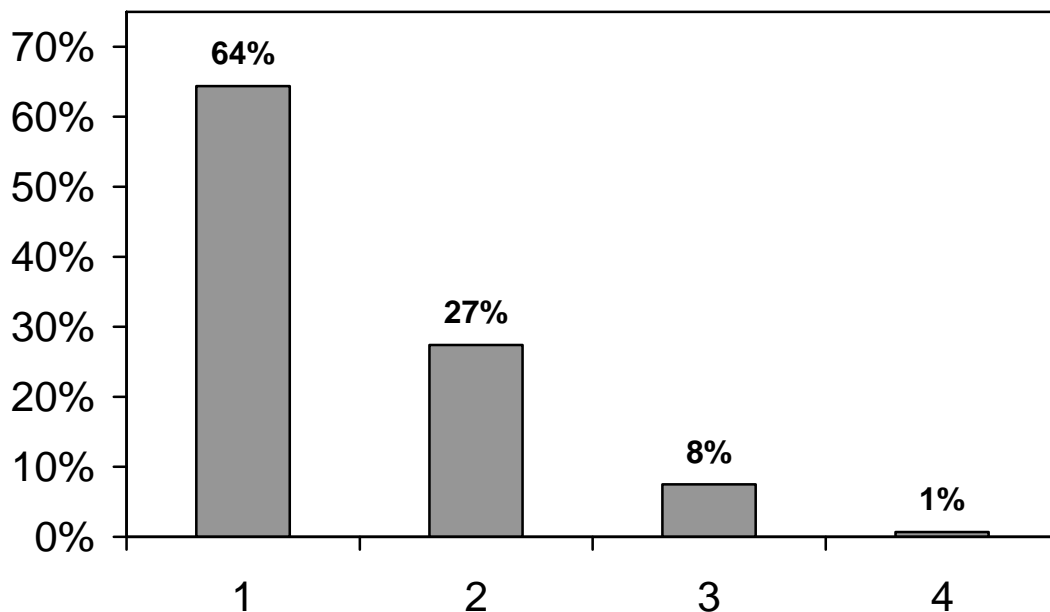


Figure 1. Number of Children with FXS in the Family

- Families live in 35 states across the United States and 1 family lives in Canada.
- More than three-fourths of the mothers (78%) and their spouses (80%) have at least some college education.
- The median family household income is between \$80,000 and \$90,000.
- The majority of mothers (73%) and their spouses (91%) work part or full time.
- Figure 2 below shows the current relationship of the biological mother and the biological father of the son or daughter with FXS. Almost three-fourths of the mothers are currently married to the biological father of their son or daughter with FXS.

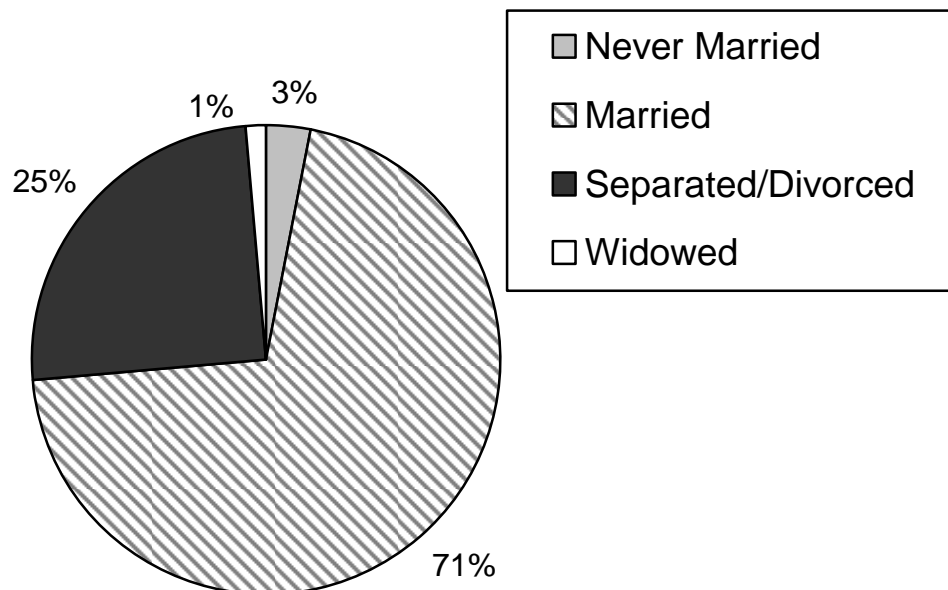


Figure 2. Current Relationship between Biological Parents



### Who are the adolescents and adults with FXS in our study?

- Fourth-fifths (81%) of the adolescents and adults are male and one-fifth (19%) are female.
- They ranged in age from 12 to 49 years. Nearly two-thirds (63%) of the individuals are adolescents (aged 21 years or younger), while one-third (36%) are adults.
- Almost half of the females (45.6%) and more than fourth-fifths (88.2%) of the males had a diagnosis of intellectual disability.
- Two-fifths (41.9%) of the adolescents and adults were rated as being in “excellent” health.
- The average age at the time they received the FXS diagnosis was between 6 and 7 years, and ranged from birth to age 46 years. Figure 3 below shows the percentage of individuals who were diagnosed with FXS at each of the age groupings.

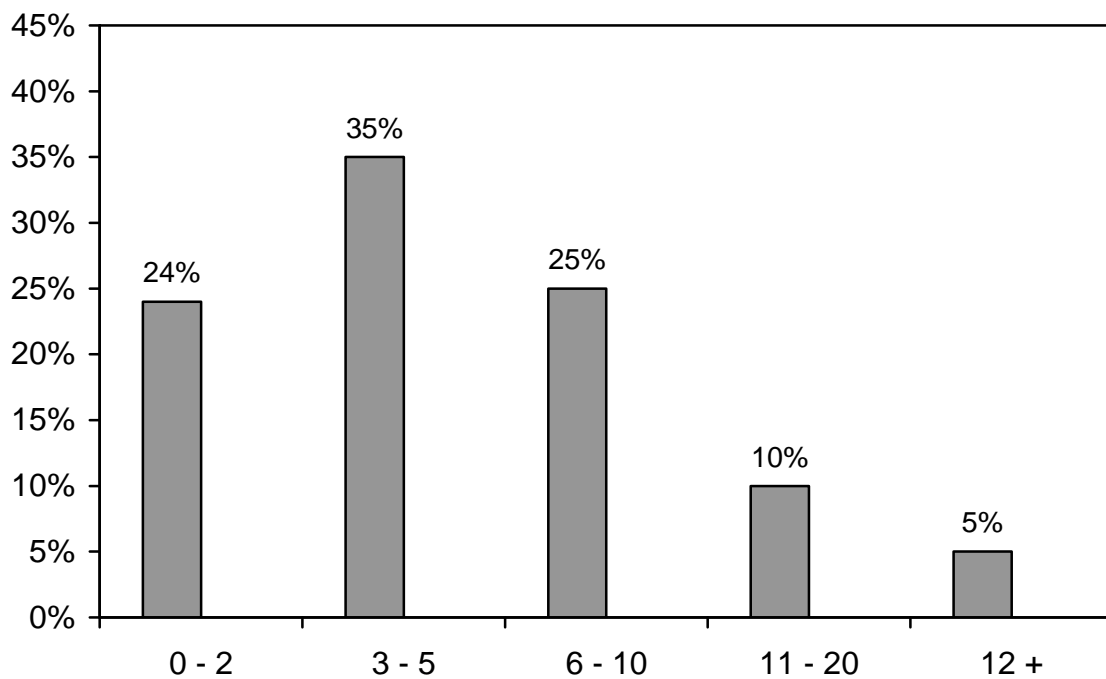


Figure 3. Age when Diagnosed with FXS

- The majority (88%) of the adolescents and adults currently live with their parents. The remaining individuals live in independent or semi-independent settings, group homes, college dorms, other residential settings for adults with disabilities, or with other relatives.
- One-third (32.3%) of the adolescents and adults had a co-occurring diagnosis of an autism spectrum disorder.

### **Section III: Comments from Mothers**

The comments made about *daughters* with FXS are in *Italics*. Comments made about sons with FXS are in regular type.

#### **Pathways to a Diagnosis of FXS**

...I read a magazine article about fragile X syndrome. I was the one that approached my physician. After I read it, I really felt that's probably what [my son] had. He was diagnosed at age 17.

*...My sister had her son tested and the results came back fragile X. She said that she was almost positive that that my children also had it.*

...He was diagnosed with autism when he was two, and within the last two years, we did a test for fragile X. He is a full carrier along with four other brothers and sisters.

...My dad got ill, he was 83 years old, and suddenly took a bad turn and was showing signs of dementia. My father's doctor did lots of testing [and found out it was Fragile X Tremor/Ataxia Syndrome]. Then, they suggested that we do a fragile X test on my kids, and that's when we found out that my kids had fragile X. And my sisters, all three of us, tested as carriers.

...My son was given the karotyping the old way. They have a history of false negatives, and unfortunately we were one of those families that got a false negative. So we went on our merry way after being told that our son didn't have fragile X. It wasn't until two years later, when my cousin's son was diagnosed with fragile X that we went back to our geneticist.

*...No doctors understood what was going on with her. They were puzzled. They ran all types of tests, but they were not getting it. Eventually, my mom was at a conference and there was a genetic doctor speaking about fragile X syndrome. We went to our doctors and all three of us - me, my mother and my daughter - found out that we had it.*

...I noticed that my son wasn't talking and I kept telling the pediatrician and he kept telling me [not to worry]. I finally asked them to evaluate my son's skills to see if he was behind. Finally, they helped me get in with a pediatric neurologist. As soon as I

went in, the pediatric neurologist asked me if I'd ever been tested for fragile X. I turned my head, I'd never even heard of it.

*...[My daughter] was first given a diagnosis of fragile X when she was 22-years-old and pregnant with her own son. I'm glad I didn't know that she had fragile X until she was an adult because I always expected her to do whatever she was supposed to do. And I think those expectations were OK.*

...I was always just looking for an answer. It wasn't until my cousin had the son with fragile X that we got the kids tested. So, it wasn't until my son was 7 [years old] that he got the diagnosis.

...They bounced us from doctor to doctor, and at one point, they were treating him for ADHD. Around age 7 they did some genetic testing, and found the fragile X. Then of course they had no idea what fragile X was, and they continued to treat him for ADHD.

...He was tested at age 4 years, and you have to realize that that was 1987, so that was the chromosome test. [It was negative.] Then, when he was about 8, I remember going to a meeting at his school, at the parent support group meeting. There was a father there who said his son had fragile X, and he had a fragile X check list. By that time it was a DNA test so that did come out [positive].

*...She was 12 [years old] when diagnosed. By sheer luck we happened to meet a stranger on the metro train. I sat next to him for 10 minutes, and was unloading about my daughter's problems. He said the he knew of a doctor who could help. He wrote down the name of a geneticist [which later led to the diagnosis of fragile X]. That stranger was our angel.*

...They basically assessed that he had global developmental delays. That was the first official diagnosis. We pushed to find out if there was anything else wrong and eventually when he was 14 months, we got the genetic diagnosis of fragile X.

...My son wasn't diagnosed until he was 42 years old. A [parent] at his baseball game came up to me and said I think your son has fragile X. That is when we got the testing done.

...At age 2 he was identified as speech impaired, but he wasn't diagnosed with fragile X until he was 9. We had several diagnoses before then, we had hyperactive, autistic, and oppositional defiant disorder.

## Understanding and Coming to Terms with the FXS

...Nobody [at the clinic] was a clinician working with kids with fragile X. So they basically said, “okay he’s got fragile X, you have to become experts on fragile X now and teach everybody how to work with him.”

...[The doctors] started talking about fragile X, and telling me it’s the X chromosome that I gave my son. They looked at me and said, “are you sure you’re OK? We’re not trying to blame you.” I said, “You don’t know how I’ve prayed for this day. You’ve told me what is wrong with my son. I’m just so thankful.” I was fine with the diagnosis because I knew there was nothing I did. I did not do it intentionally. I just had to deal with it. At least now I understood.

...At the time he was diagnosed, we did not have a pediatrician that really knew anything about fragile X. So I found a book in our library all about fragile X, and I educated myself on it and I tried to educate the teachers.

...It was frustrating in the beginning not having a diagnosis, because there were a lot of suggestions by other people of what it might be, or what I might have done, or what I should have done. So, it was a relief to get a diagnosis.

*...The difficulty in diagnosing girls with fragile X put my daughter at a disadvantage. She probably would have benefited from early intervention, but we didn’t know the diagnosis. So if anything comes with research, let doctors know that girls can have fragile X and not present with mental retardation.*

...I think that in general with fragile X it’s very important that you just keep pushing to get the information out there because it took, I don’t even know how long, before I even knew anyone else with it. It was like we were always alone, because the doctors didn’t know anything about [fragile X] and you didn’t know any other people that had children with it.

...I have the three boys, all of them have fragile X. There are a lot of similarities, but there are so many differences between them. I think that’s pretty amazing that they can all have the same thing and yet have different personalities, different challenges.

...Learning that your child has fragile X is helpful in some ways. On the other hand, I had six years where I did not see my child through the lens of disability. Once they’re disabled, you always associate this behavior with the disability.

...It's been very hard to know that I passed on this gene. Everybody says you shouldn't feel that way, and I say okay I won't. But it makes a difference. Had I known, I probably wouldn't have had children. But thank God I didn't know because they're all a blessing.

...A lot of people know what autism is, and that's the only way I know how to explain it to people. I wish there was more awareness about fragile X.

*...It was hard not knowing her diagnosis, and feeling guilty thinking that I had done something wrong to make her this way. Finding out that she has fragile X has helped give me a reason for her disabilities.*

...When he was diagnosed, it was a big blow to me because I felt like I had given [fragile X] to him since it was passed on through me.

...Very shortly into raising [my son] I discovered I had to throw out all the baby books that had anything to do with developmental norms. I have had to rely on my own instincts.

...I had a really hard time understanding my sons before I knew it was fragile X syndrome. But now, it's almost harder because it's like I'm at a dead end. Before, I was reaching for things - for [my sons] to reach different levels and learn new things. But now, I think that maybe this is as much as they can learn.

...On one hand it was such a relief to know why, and then on the other hand it was so overwhelming. I went through a period of anger and denial, the whole thing, until you get to acceptance. Even when you get to acceptance, there comes certain situations when you're like this isn't fair, and you go through all the emotions again.

*...Before my daughter was diagnosed, I was told I was too hard on her, I was told I was too soft on her, I was told that I didn't breastfeed her long enough, and that I [shouldn't have] had soda pop during pregnancy. I had a lot of guilt. Once she was diagnosed I was almost pleased because I realized it wasn't my parenting. I was the model mom doing everything, and I didn't understand what was wrong.*

## My Son or Daughter's Strengths

...His personality is his greatest strength. He wakes up with a smile in the morning and he goes to bed with one.

...He's well known and liked in the community, in our church, in school, and in the Boys and Girls Club. He seems to know more people than me. On the street, people will say "Hey." And I'm "Who is that? Who is this one? Where you know that one from?"

...He is quite a character. He can be pretty funny. Out of the blue, things will come out of his mouth that will blow you away, and you'll just die laughing because he's just so appropriate and at the right moment. I mean it's just hilarious.

...He's kind, he's generous, he's handsome, and he is a friend to everybody. For example in the morning in his [residential home for adults with disabilities] there are about twenty boys, and some of them are not as high functioning as [my son], and they can't tie their shoes. So they just come in [my son's] room, sit down on his bed, put their feet up, and he ties their shoes for them every morning.

...He would have probably been a politician if he had not had fragile X. Anyone that gives him a half of a chance ends up loving him

*...She's really good on the computer. I'll never understand that in a million years. Seriously, if I need something on the computer I ask her!*

...He's on Facebook and has a hundred and ninety-one friends right now. And he's counting to when he gets to two hundred. He has very limited language skills, but is able to communicate on Facebook with simple language and words.

...He wins people's hearts by remembering names. He has a great meet-and-greet. He'll go up to them, shake their hand, and call them by their name.

...Every day someone says to me how much fun he is. He brightens up their life. The bus driver, the school nurse, whomever. He's just full of fun and he always wants everyone to be happy.

...One of the happiest kids on the planet. He literally will go around saying things like "It's good to be me." And "I love this town." And "Life is great." "Today is the best day of my life." Most kids are so weighed down by the need to please their

parents, the need to do well in school, the need to make the baseball team, the need to compete with their sister, the need to make sure that their best friend is paying enough attention to them- that they really have a lot of concerns on their mind. Our son seems to be so completely removed from any of those things, that it just allows him to enjoy life.

*...She's very loving, very helpful, good relationship with her brother who also has fragile X syndrome. My son is more severe. She has such patience with him and helps me take care of him. She can cook very well and even though she doesn't want kids she's great with them and they love her. And she's happy.*

...He is a wonderful young man. He's very caring. He notices the feelings of other people. Recognizes when there's something wrong, and loves to hug people and ask if they're okay.

...He is just so family oriented. He is nuts about his family, extended family, aunts, uncles, grandparents, he just feels a real sense of caring for the family.

*...She wants to please and do what's right. I think that's going to help her too, she's going follow the rules, and make sure she's there on time and she's going to try her best.*

...I think his compassion for people, his innocence, his sense of humor, his overall attitude [are his greatest strengths]. When he's asked to do something, he'll just say "I'll try to do it."

...He seems to draw people to him. It's quite amazing how he can just walk in and start talking to people, and they just respond to him. It's just amazing how he does that.

...He's a great kid. He's funny. He has a great sense of humor which is a nice attribute because people relate to him and love him.

...He's very, very helpful. He was voted most helpful in his second grade class by his classmates and [his helpfulness] persists to this day.

...He has a great sense of humor and is very social. He is also very loving and affectionate. He can tell when I'm not feeling well and he tries to be caring and helpful.



## **My Son or Daughter's Challenges**

...His ability to fit in with his peers is a challenge. He's always wanted a close buddy. He had one at school, and then that just ended. He doesn't get to hang out with guys. They don't call him and don't invite him to go to the mall or the movies or stuff like that.

*...She really struggles with abstract concepts. Anything that isn't clearly obvious and easy to understand, she doesn't get.*

...His greatest challenges are that he is non-verbal, he's not potty trained, and he requires a one-on-one aide in all settings.

...A lot of his behaviors, like whacking people or wetting himself, keep him from doing things in the community.

...He can only say one and two word phrases to express his wants or needs. He gets aggressive and pulls my hair if he is frustrated and cannot communicate what he wants. He also wants to eat all day long, and I have to keep him from doing that. He's like a three year old who is told "no", but he is six foot three and weighs a hundred and ninety-some pounds.

...He wants to learn how to drive. He talks about living on his own. He talks about things that most everyday teenagers talk about. He gets frustrated because he recognizes that he might not be able to do those things.

...He does not have good language skills and other people have trouble understanding him. I know that he would say more if he weren't so frustrated that people don't understand him.

*...She has a lot of trouble with inattention. She doesn't like to pay attention and will come home from school and not know what she's supposed to do for homework.*

...He's very destructive. He likes to break things and rip up shirts and things, which makes it hard to go out in public or take him places.

...His obsessive behaviors are challenging. Having to have the same sort of clothes on all the time, or having the same color sweat pants.

...He'll say "I can stay here alone. I can stay by myself." He doesn't quite understand that "Yes. You're a grown man. Yes. You're twenty-two. But you're not like the other kids."

*...Most people think before they speak. She thinks as she speaks.*

...He really struggles because he wants a girlfriend, he wants to date, and he wants to get married, and he wants to do all of the things that a boy his age should do. But, he knows that he probably won't do them and that hurts him.

...Money for him is horrible. He can buy something for a dollar fifty, give [the clerk] a ten, and never expect change. That is a real concern for me especially as he gets older and is out in the real world.

...He is nonverbal and can't express when he's upset. I know he gets upset and has blow-ups and it's ninety percent because he can't express himself.

...Not knowing how to read people in social situations and being taken advantage of [are his greatest challenges]. We don't let him get on the computer, which would be a wonderful tool for him, because he has misused it. It's very scary for us, he gets pulled into things very easily and it's scary in this world.

...He does not take initiative to interact with friends. We try to schedule outings with friends, but he will not call them on his own. Consequently, he is isolated from people outside of his family.

*...We always have to push her to go do something, and afterwards she'll enjoy it. But it's just that constant pushing her to go do something.*

...He has trouble with cognitive skills like math, reading, and sequencing. If he tells a story, it'll always be yesterday even though it was really a week, a month, or years ago.

*...She's very sensitive to feeling hurt or lonely or rejected at school. If she thinks that she doesn't understand something, she's very self-conscious about speaking up. She's not very confident at all.*

...He has the standard teenage defiance. Yet, he still needs me. It's like, "Leave me alone. Leave me alone. No. Please give me a hug."

## **Impact on Siblings without FXS**

...I think it has been a huge responsibility for her because all of her life she's been, more or less, watchful over her brother [with fragile X].

...I feel like they've had to make a lot of sacrifices because growing up, everything had to revolve around their brother with fragile X and what was best for him.

*...It's hard for him, especially when [his sister with fragile X] is going through her behavior issues, because she would hit him and say things to him. He just tolerates it.*

...I think it's given her confidence. She's always been on the shy side. But when she saw that her brother with fragile X didn't have inhibitions, it helped her. She saw that her brother could go to the sidelines of the football game and jump around, not acting in a typical way, and people liked that. It helped her get rid of some of her inhibitions.

...It was hard for her to have friends over because she didn't know whether her brother [with fragile X] would be having a good or bad day.

*...I think she feels like she doesn't get enough of my time and attention. She may feel like she is pushed aside.*

...She has an unusually high tolerance and acceptance of people who are different. She is a very giving and loving person, because of her brother with fragile X.

...I think it makes him lonely. His brothers with fragile X don't do the things that he likes to do like sports and play board games.

...She feels responsible for her brother, even though we have tried very hard not to make her responsible for him. I think that there's probably some survivor guilt of the fact that she doesn't have fragile X, isn't even a carrier, and her brother is so affected

...I think it was sometimes embarrassing to have friends over, so he didn't have friends over.

...If she and I are going somewhere and her brother [with fragile X] can't go, she doesn't want to go. She wants her brother to be with her.

...I think it's made her sensitive to a diverse population of people. I know she cares about him, they care about each other, very much and I know it has made her sensitive to that particular population.

...My two daughters [without fragile X] are extremely close with each other. They don't ever fight. They share a room even though we have an extra room. I think, in part, that's the result of having a brother with fragile X, who has such challenging behavior.

*...I think sometimes she's embarrassed, frustrated, and sometimes she wishes that either her sister with fragile X were different or that she had a different sister.*

...She was very jealous of [her brother with fragile X] when he was very young, but now she's more protective of him. She is going to be his guardian if something happens to me and her father, and make sure that he is taken care of.

...I don't think it's different from any other sibling. They're just brothers. One would jump on top of the other and then the other would punch him and they have fights, but they get along.

...As a child, I think he felt resentful. We were always taking his brother with fragile X to therapy and here and there. It seemed like his brother with fragile X got more attention. At times, he would say "you care more about [his brother with fragile X] than me." However, as an adult, he's very supportive and helpful. If my husband and I have to go out of town he'll come home and hang out with his brother with fragile X.

...At times she felt that her brother [with fragile X] got more attention than her. She doesn't have a lot of patience with her brother and has a lot of pent-up anger. But, she's working through it.

...I think the biggest thing would be that we made my son [without fragile X] do things with his brother with fragile X when he was younger. If he was going to go do things with friends, we said "your brother [with fragile X] is going to go too." But otherwise, I don't really think it's been a problem for him.

## Parenting Struggles

...You never know what to expect each day. You never know if something is going to set [my sons] off. We try to keep everything as routine as possible. It is frustrating when after supper you want to go out for a drive or you want to run down to Walmart, but their routine is that after supper they go to bed. So they don't want to go anywhere.

...We didn't have anyone that we could leave our son with that understood how to take care of him.

...School is a struggle because it's hard to get people to understand him. Even when I think that the teachers and staff understand his disability, they don't. For example, the nurse never seems to be able to handle his anxiety attacks. She calls me instantly and says, "Maybe he should come home." She just gives up immediately and is afraid of him.

...My husband and I are hardly ever able to get away together.

*...She's very dependent on me even though she's struggling to be more independent. We fight. I would say constantly. It's been a lot of teenage stuff.*

...There is not a lot of respite care for him. Even when he was little it was hard to find somebody that could handle him or understand him, and that's been an issue all the way up to adulthood.

...We have a love-hate thing going on. He needs me, but he doesn't want to need me. I tell him to do things a certain way but I also try to back off and let him find his way. This happens with simple things such as ironing his clothes. He would walk out of the door with half of his pants crumpled and half of his pants ironed. It's very tough for me to leave him alone and let him learn his own way.

...I worry if he's going on a class trip. Is he going to get panicky, or is he going to get anxious and nervous? I feel sorry for him because I don't want the other kids to see him like that. He could care less. It's almost kind of funny that I'm the one worrying about it and he could care less.

*...Whatever works one day, the very next day you try to use the [same] calming technique and that's not going to work. So it's been very challenging to find out what's going to help her and what's not.*

...Because of [my children's] special needs, we have no privacy. With the girls in foster care and all of the people that my son has to work with, you couldn't live in more of a glass house than we live in. There are unflattering parts of me that now everyone knows exist.

...It's hard because he's a very large seventeen year old that's really mentally three. It's like having a gigantic toddler that can throw pretty massive temper tantrums and I do not have any help. I do not have anyone that will watch him. The only person that can watch him besides me is my fifteen year old daughter. Other than that, no one even wants to attempt it.

...I don't want to call it a crisis, but there was always an issue. We couldn't go on our family vacation or spend a day at the beach. Always at the back of my mind was what if this happens, or how can I quiet my son down enough to convince him to stay for the other kids.

*...She doesn't leave the house. She's home all the time, so that in itself can drive a parent crazy.*

...I call us the "fractured family" because we always go places in two cars. One parent goes with our son [without fragile X] and the other parent brings our son with fragile X later on, so that we can make a quick getaway if needed.

...Having two special needs kids, that's a huge barrier to maintaining friendships. It's like, why haven't you called so and so in six months? My kids take up so much time, and because our lives are so complicated. I really couldn't go to that Tupperware party because I had to spend three hours looking online for adult swim diapers.

...We're with him all the time. It's like having an eighteen month old child in an eighteen year old body.

...He still needs help with picking out his clothes and bathing. He can fix himself small non-cooked meals. But you still have to watch him because he's big on sweets. He would like to eat all of the cake instead of a piece of the cake.

## Experiences as a Mother

...It's been the most wonderful thing. It has also been one of the most challenging things. There have been times in my life where I've felt totally alone and totally frustrated and there have been times when it has been a real strain on the marriage. But overall I think it has made me a better person.

...It has made us into different people, people that we wouldn't necessarily have become. I think that we probably had to delve a little bit deeper into things than other families and find what's truly important and appreciate the smaller things. It's not to say that we're happier because of it, but it's not all bad.

...Once I got over the grief of losing the child that I didn't have, or thought I was going to have, then I was able to see that I had a purpose in this world. That my purpose was to have him. Once I understood that, then I felt lucky.

...It's been a wild ride. Three out of the four of my children have special needs.

...I had a very demanding job. But in addition, my second job was coming home, trying to do the therapies with my son, and [my] third job, late into the night, was fighting with insurance companies, trying to get them to pay for the therapies.

*...I think it has made me a much better person. It forces you to know that your life isn't just about you, which is how it should be. It forces you to be patient. You learn to let go of what people think.*

...I always feel like I'm running a marathon and I can't slow down. I'm kind of beat up, kind of worn down, on the bad side, and kind of feel like I've been drained.

...It has been challenging, sometimes frustrating, depressing at times. And very rewarding, probably more rewarding than anything.

...I get sad when other people don't see what I see in [my son], but I don't get sad about who he is, or what he will or won't be able to do. I just get sad for limitations that other people might place on him.

...When he was first diagnosed, I fell apart and I didn't know what that meant for his life. Through the years, as I learned and as he grew and I grew, I realized that he helped me be who I am, and I'm kind of happy with who I am.

...It has beautiful rewards, but it also has its challenges and frustrations. But I'm probably a 500% better person than I would have been if I hadn't had my son.

...I was never a mom. I was an advocate, I was a social worker, I was the doctor, I was the pediatrician, I was the case worker, I was the DVR representative, I was you know everything.

*...I kind of feel like I live in a different world. The friends we have are very far and few between because we just don't live in the same world.*

...Raising my kids, I've learned a lot from them. I've grown a lot from them. I feel like I'm a more empathetic individual for having a child with special needs.

...It's a trade off of problems. I don't have the information that people with normal kids have, like books [on normal development]. But I won't have to worry about drugs, or driving, or things that parents with normal kids have to worry about.

...I spent many years feeling guilty, thinking that it was my fault. I've been through a lot of counseling. I was jealous of my sisters and their normal children.

...I'm tired. I'm definitely tired a lot. I go through phases. Sometimes I feel very blessed and lucky. And I love my family. I love that we are different. I love the way we are structured.

...For all the hard times and all the really rough times, I think all in all we are better people for it. I would never ask for it, don't get me wrong, I would never ask for us to be the people with the child with a disability.

...I would say fragile X has probably made us more spiritual, and we've had to have faith to get through certain situations.

...Our message to our kids is "oops sorry guys, but [fragile X] is what we have." That's just our genetic makeup, so we're very accepting of what our genetic makeup is. We try to work with it, you know, in a cooperative way.

*...I think having a kid with a disability makes you appreciate the little things in life and makes you not take everything for granted.*

...It's always been like an uphill battle, a challenge. There are of course minutes – moments - of lots of laughter and joy.



## Concerns and Plans for the Future

...I become more concerned about my age and what his future is going to be like without me, when there comes a time when I can't physically take care of him or if I were to pass away so that bothers me. I think about that a lot.

...We're not sure if he'll ever move out, if he'll always live with us. If he does move out I'm thinking, wow what's that going to be like? Can I trust somebody else to take care of him? How could I ever have him move out and let somebody else take care of him?

...I am concerned about his future because he is unable to handle money, make important decisions and he is dependent on his family for guidance, finances, and transportation.

...Sometimes, not often, I struggle with feeling guilty because I wish I had privacy with my husband. I just wish there were more options for places for my son to live and be cared [during his adulthood].

*...I think that being independent after high school is going to be a huge challenge for her. I don't know if that's going to happen. And it's kind of nerve-wracking. She won't even talk about it. So she'll probably be living with me forever. And she's capable, you know, she just isn't willing.*

...My biggest concern is the future. Who will take care of him when something happens to us? Will he have more challenges as he gets older or will he stay about the same? What will he do with his free time when he can no longer participate in Special Olympics? Will there be medical challenges? Because he is so close to our immediate family, I worry that he will regress if he has to move to a new setting.

...It gets harder as he gets older, because he gets more isolated and there's more responsibility on us to get him out to get him doing things. That's hard because sometimes you don't see an end to that. We're just always going to be responsible for him and that's hard.

...When he was younger, he always loved school, he always had really good relationships with his teachers and we loved that. So it's hard when you move on from there to try and find a community for him.

*...She's very trusting, which sometimes is a problem because she's in an adult residential setting and workshop, and sometimes the other people take advantage of her. I'm worried about her being taken advantage of in the future, when I can't be there.*

...The future is a challenge. Who will take care of him if something happens to us? The burden that might be placed on someone else. The fact that he could be so easily be taken advantage of. The fact that to the general public, there's really no obvious sign that he has a disability.

...I know I'll never have empty nest syndrome. Now that I'm at the age where my friends are getting to that stage of their lives, I don't feel like I've been cheated, but I feel like I have to have a different lifestyle. It's not good but it's not bad. I'm limited to things.

...You go from hovering over them at every second to thinking "okay, it's getting a little bit easier now [as son ages into adulthood.]" But things are getting more interesting now because you're planning for the future. So there's a lot more worries about what the future holds.

...I worry a lot about what's going to happen to him when I'm not around. I've kept him at home with me because I'm not satisfied with a lot of the programs that are available out there. At seventeen, he's in what we call the "gray area" of getting services. There's a lot out there for the younger children or for adults, but he's stuck in that gray area where there's really not anything.

*...I'm finally able to say "When I'm not here she's going to be okay on her own." Granted I'll still have her brother or someone looking after her.*

...I don't ever see him living out of my house. We bought a home where we can make sort of a little apartment for him when he's older, but I don't know how that's going to work.

...I am so relieved that he is happy living in his own apartment. They clean. They do what young people do. The challenge continues to be for him to get a job, which he hasn't been able to get yet.

...I think he has a good future ahead of him. We plan to always have him stay with us. Right now I'm developing a business so that he can have a job and work with friends who are his age.