

Waisman Center
University of Wisconsin – Madison

Advancing knowledge of human development, developmental disabilities, and neurodegenerative diseases.

Study on Family Adaptation to Fragile X Syndrome



Report 8: Experiences of Premutation Mothers and their Siblings

- Principal Investigators -

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If you would like to read other family reports or research papers from our research group, please visit our study website:

<https://family.waisman.wisc.edu/fxs-fmr1-associated-conditions/>

For more information on FXS and related disorders, please visit the NFXF website: <https://fragilex.org/understanding-fragile-x/fragile-x-101/>

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Section I: Introduction

For over a decade we have been conducting a nationwide study entitled *Family Adaptation to Fragile X Syndrome*, funded by the National Institutes of Health (NIH). This study's focus is on families of adolescents and adults with fragile X syndrome (FXS) and is the largest and longest-lasting study of its kind. Over time we have asked questions about the physical health, functioning, and emotional well-being of adolescent and adult children with fragile X syndrome as well as their mothers with the premutation.

We have learned a great deal about family adaptation to FXS to date. However, there are additional perspectives that are important when studying FXS in the family context, and one of these is the focus of this report. Here we report on information shared with us not only by the premutation mothers who have been participating in our longitudinal study, but also their biological brothers and sisters. In other words, for this report we are studying one generation – the premutation mothers and their siblings.

All of the premutation mothers in our Family Adaptation study have children with FXS, but their siblings vary in whether they also have the premutation, whether they have children, and whether their children have been diagnosed with FXS. As you know, the gene that causes FXS is transmitted throughout a family. In some extended families, only a few people have the premutation or FXS, while in other families many have these diagnoses. A goal of our research is to increase our understanding of the overall well-being of the entire family, including those who do not have a child with FXS.

This report is divided into two main sections. First, we describe the “sibship” as a whole – based on information provided by the premutation mothers who are participants in our study. The term “sibship” means **a group of brothers and sisters born of the same parents**. We report on factors associated with the sibship including age, birth order, number of siblings, sex, frequency with which siblings are in contact, and feelings of closeness.

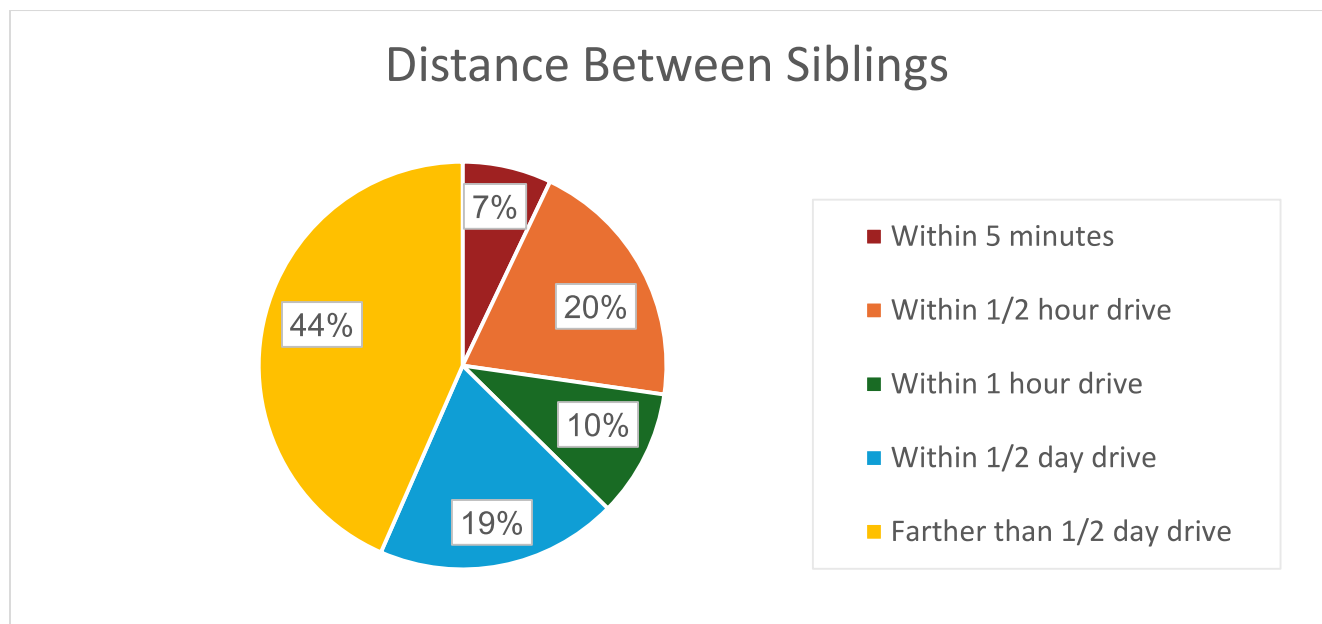
Next, we report on findings from our “Sibling Study,” which we conducted in 2019. For the Sibling Study, we started with the premutation mothers in our longitudinal study, and invited their siblings to participate. Our goal was to learn more about the family context of the *FMR1* gene within a single generation. The sibling relationship is the longest-lasting family tie, so studying this relationship provides a critically important window into family life. To preserve the anonymity of the siblings, only their first and last initials were recorded.

Section II: Describing the “Sibship” in Participating Families

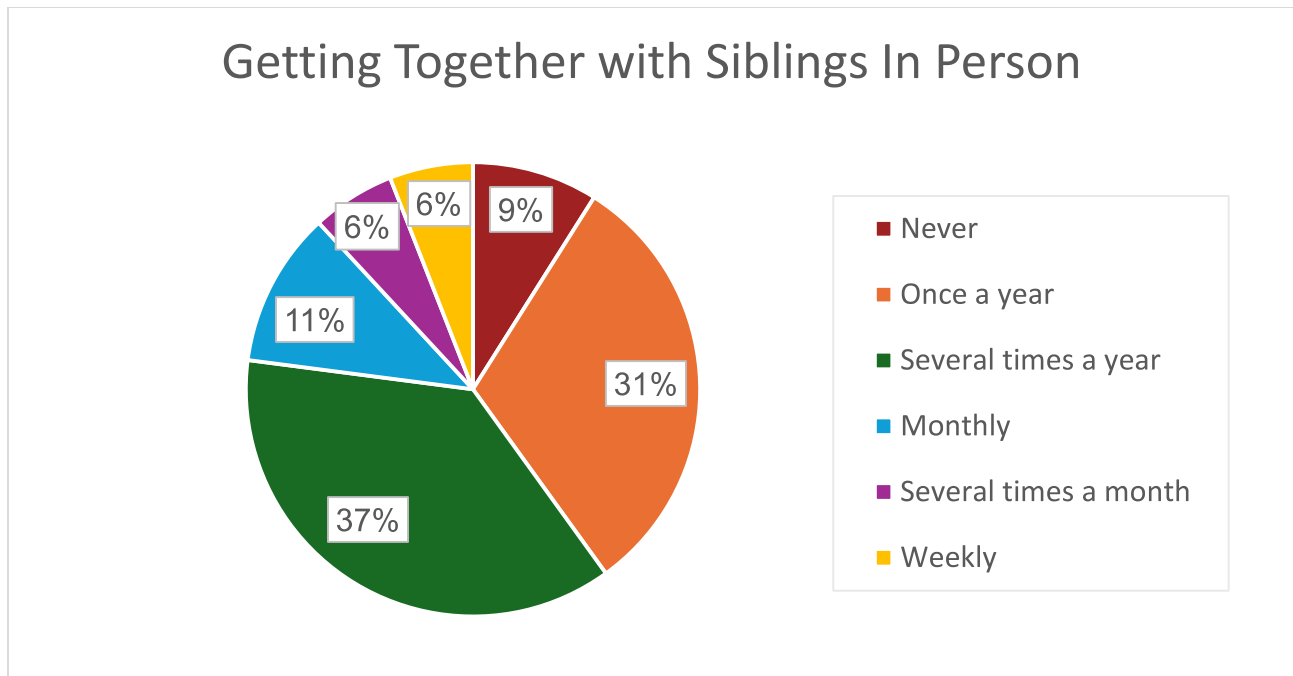
What We Learned About the “Sibship”

In total, the 109 premutation mothers who were participating in our study in 2019 had 253 biological siblings. Almost half were brothers (52.2%) and half were sisters (47.8%). Family size varied from 35% of families where there was only one sibling, to nearly 10% families where there were more than five siblings, with an average of 2.3 siblings of the premutation mother per family.

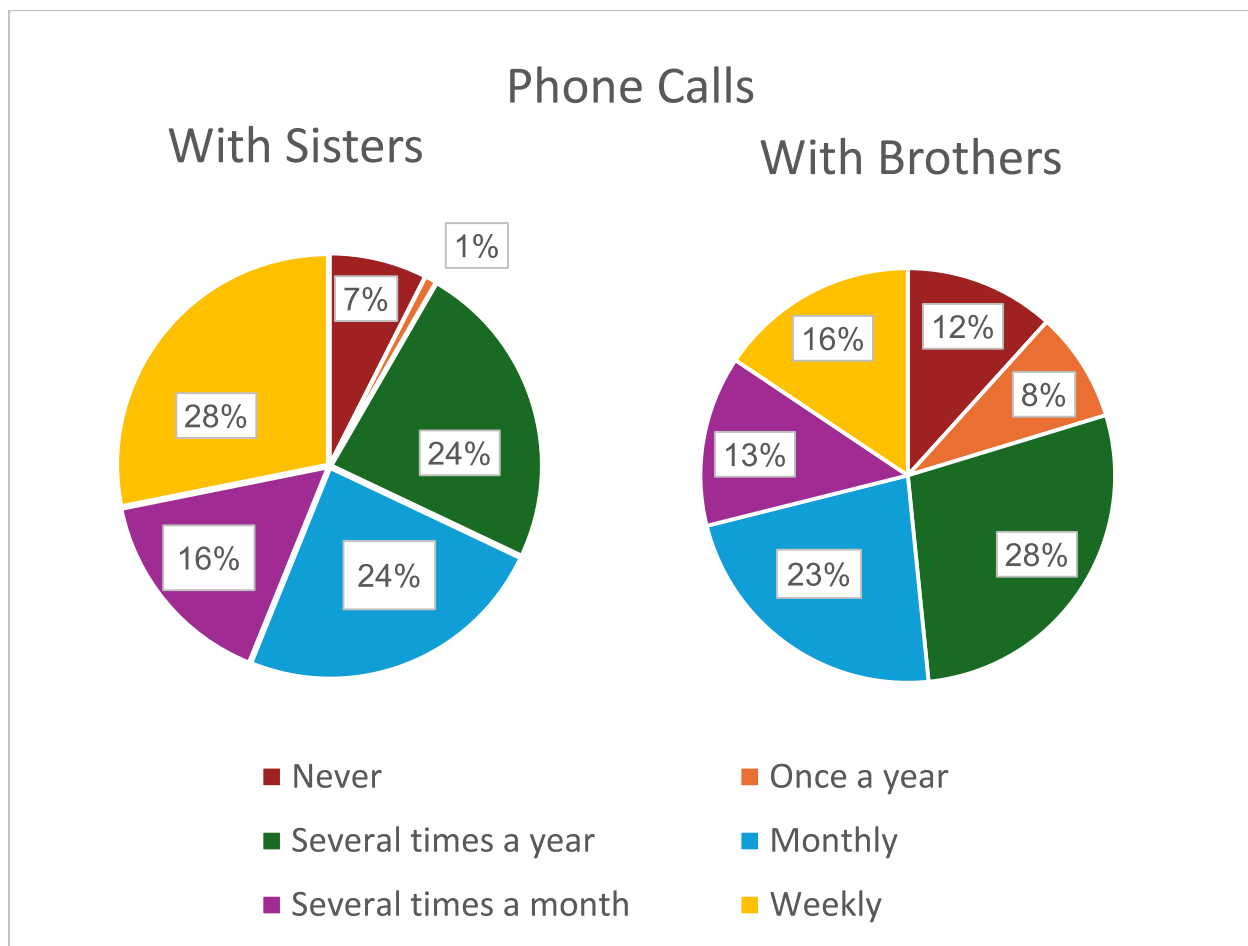
There was a great deal of variability in the **distance between where siblings lived relative to each other**. 27% lived close by -- less than a half-hour's drive away from their sister with the premutation who was participating in our longitudinal study. But almost half (44%) lived more than a half-day's drive away. There were no differences between brothers and sisters in these residential patterns.



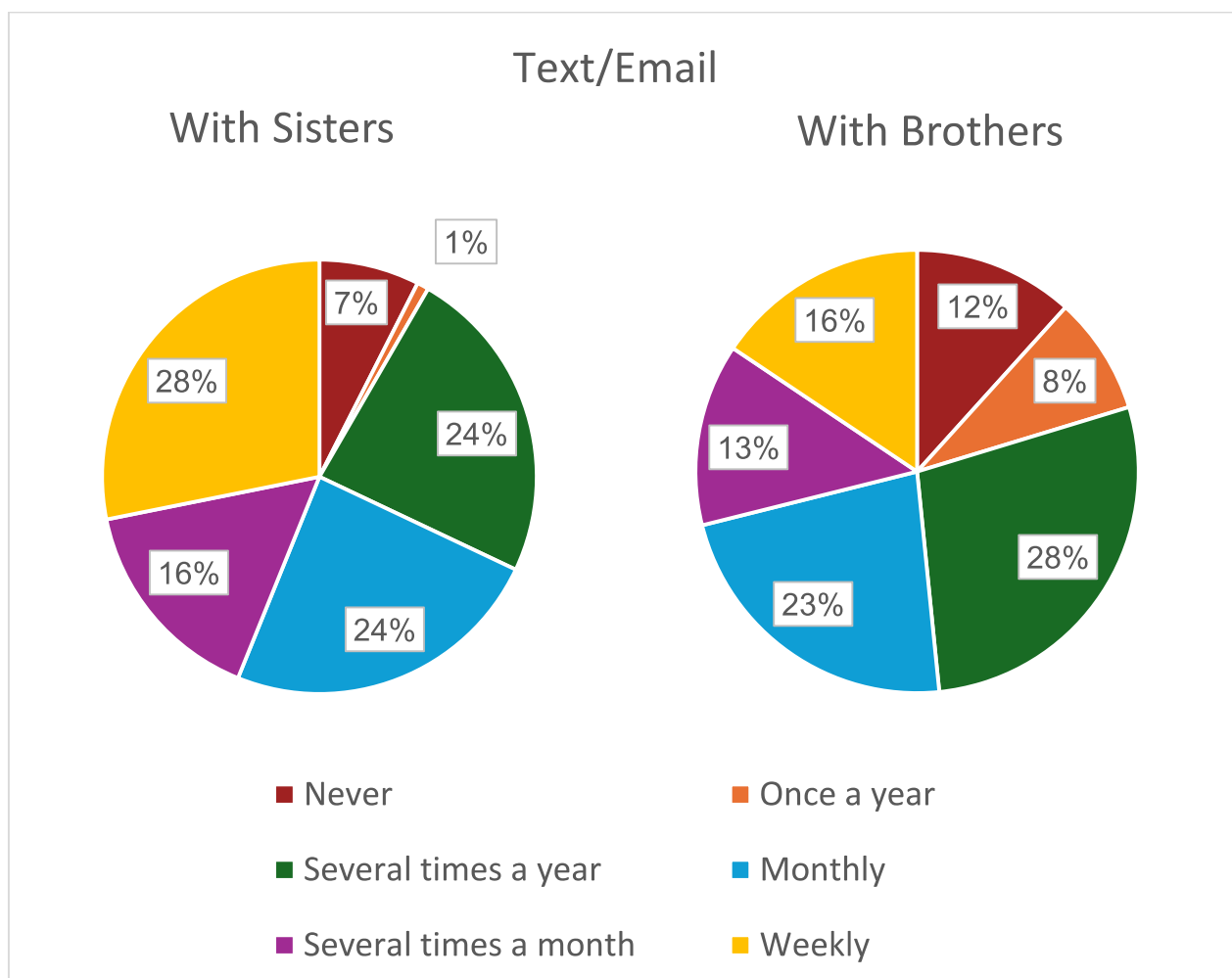
In terms of **getting together with siblings in person**, there also was a great deal of variability. 6% had in-person contact with a sibling at least weekly, while others reported much less frequent in-person contact. 31% reported getting together in person once a year or less often, and 9% never visited with their siblings in person. Again, there were no differences between brothers and sisters in these patterns.



However, there were distinct differences in **frequency of phone calls** exchanged between the premutation mothers and their sisters versus their brothers. 28% had weekly or more frequent phone contact with their sisters, while about half as many (16%) had that frequent phone contact with their brothers.



In some families, **digital forms of communication** were more common (texting or email) than phone calls or in-person contact. 28% of the premutation mothers communicated via text or email with their sisters weekly, whereas 12% of them never texted or emailed with their brothers.



Not shown in a figure, in addition to geographic distance and frequency of contact among siblings, premutation mothers provided information about their **emotional closeness** with their siblings. Overall, premutation mothers were quite close to their brothers and sisters, but were more likely to feel “very close” with their sisters (29.1%) than their brothers (16.4%).

Summary. The premutation mothers in our study have many types of exchanges of support with their brothers and sisters including contact in person or by phone or digitally, emotional closeness, and a shared family experience. But in general, in these families, as in many US families, the sister-sister bond appears to be closer than the sister-brother bond.

Behind the numbers and percentages reported here, are the perspectives of the siblings in the family. In the remainder of this report, we will let them share their observations and feelings in their own words.

Section III: The Perspectives of Siblings

With permission from the mothers, we invited each of their siblings to participate in an interview and complete a questionnaire in order to provide a deeper understanding of the perspectives of the siblings within a family. Almost half (44.2%) of the mothers had at least one sibling participate in the Sibling Study. There were no statistically significant differences between mothers who had a sibling participant and mothers who did not other than their number of siblings. Our goal was to learn more about the experiences of the brothers and sisters in their own words in order to increase our understanding of the impact of FXS on the entire family.

As part of this Sibling Study, we asked each sibling to reflect on the impact of FXS on their family, resulting in discussion of:

- Family ties
- Family planning decisions
- Feelings of love, pride, and gratitude
- Understanding of disability
- Thoughts about the future

Each of these topics is explored below, with selected quotes from siblings. We end with siblings' thoughts about the need for **research, education, and awareness about the *FMR1* gene** in order to expand understanding of the family-wide impact of the premutation and FXS.

We acknowledge in advance that siblings provided many more quotes than we have room to include here, and since some included quotes were very long, we occasionally shortened them. We tried to select the quotes that were representative of the larger set of comments, and we hope we were able to capture the most important messages of the siblings in these unique families. Identifying information has been removed to protect privacy and confidentiality. Most importantly, these are the siblings' perspectives, in their own words, and we are extremely grateful to them for sharing their thoughts and feelings with us.

FAMILY TIES

“In terms of my family, the experience we've had with my nephew, who has Fragile X, has been difficult at times, has brought us closer at times, and has made all of us feel grateful.”

“I think that my children, and me too, realize how difficult my sisters' lives have been with having a child with Fragile X, and I think it has made our family stronger as far as looking out for each other and helping each other and just basically being there for each other in all kinds of situations, not just with Fragile X, but with other situations you know such as normal family situations. I think it has helped our family to be really close.”

“I try to be as supportive as I can, and we all try to support as much as we can, but otherwise we have a pretty close family and I feel blessed.”

“I see the impact that this has had. From my point of my view, it has made me closer to my nephews. I want to do for them as much as I can to help them out. This has been a life changing experience I know for all of us in the family to have the two boys like this.”

FAMILY PLANNING DECISIONS

“I would say it has been a significant effect on all of us, especially for me and my husband and making the choice for us having another child, and the implications of that.”

“By me getting tested, we could figure out where this came from, we figured out it came from my dad, so we all [female siblings] had it. His diagnosis gave me the information I needed to know.”

“It's one reason that we've only had one child and that's the way it works out, you know. You play the hand that you're dealt and you make the best of life and things go on. We've never had any regrets not having other children. There's a lot of people that have things a lot worse than we do, or my sisters do, so I guess that's it.”

“I can remember when the boys were first diagnosed, and how my sister encouraged all of my cousins to get a DNA test to see if they were potential carriers. And, how she stressed the importance of that. I know my brother and his wife were tested before they decided to have children.”

FEELINGS OF LOVE, PRIDE, AND GRATITUDE

“As far as Fragile X goes with the family, with my nephew, he's doing wonderful. My sister is such a great advocate for him and he's come a long way over the years. And obviously life has been challenging but we all love him and he loves us and he's thriving. So, all in all, I have a lot to be thankful for.”

“My sister and her husband have made it easy for family members to be supportive of them and of their son with FXS. I cannot say enough of how proud I am of my sister and her husband for the way they are living their lives and the way they are raising their son. We're proud of their son with FXS and his gentle heart and his loving nature, and it's a pleasure to have him as a nephew. Every accomplishment he makes, we are just so proud of him, and he is a joy, an absolute joy.”

“My sister has been an unbelievable advocate for her son. She has just been exceptional in the way she has chosen to fight for him to give him the care and love and support he needs. We have seen the benefits of her passion in getting him the education he deserves. And, we are really all of us are in awe in what she has done in her experience with raising her son.”

“If it weren't for them, we as a family would know nothing about Fragile X. She has done everything in her power to educate us and she does everything for her son as far as trying to make him be accepted and live a normal life.”

“My sister and her husband, were outstanding parents to their son with Fragile X. They were both very loving and caring and worked hard to make sure he had a good life.”

“I will say that my sister has done a good job with her son and given him all the help and support he has needed. He is a really good guy, and I am really impressed with how he has come through this, and it has given me a lot of respect for Fragile X people.”

UNDERSTANDING OF DISABILITY

“Well, it opened our eyes to a lot of things that we were never aware of before about special needs and lack of help and assistance in the world with those who have special needs”.

“It has affected the family I think in a positive way. It’s affected the family in that we are more aware of disabilities, mental disabilities with people, and are more accepting and patient with them. It has been very eye-opening and has made my children more compassionate and more willing to stick up for other people.”

“I believe it has made me and made us more aware of developmental needs and just being more compassionate and patient with people who have those issues. Just learning more about how genetic issues run within a family, how that happens. We are more aware, understanding, and patient.”

“We have a lot more empathy for people with developmental disabilities. Ever since my older sister's son was born, we have been very supportive. And, we try to be very understanding of anybody who's in that position, whether they be the caregiver or the parent or the child themselves.”

THOUGHTS ABOUT THE FUTURE

“We want to position all of the family members that have Fragile X for life beyond when my generation passes away. That’s the biggest focus of the family right now. My sister and brother-in-law are in the process of trying to, create an environment that their son who has Fragile X will live nearby on his own with appropriate support mechanisms, and for the cousins that also have Fragile X, everyone’s trying to do the same thing.”

“My goal for five years from now is to be able to provide a multi-unit, small apartment building for people with Fragile X in their family so that the child who has it will have a living environment after their parents pass away. That’s my goal, is to be able to provide that through a foundation, which I’m working on.”

“But certainly, as we age and become in need of care ourselves, there’s the “what if” scenario. I think that's the space we're getting into now as we age. Because of services and things like that, I'm seeing more concern as my sister and her husband continue to age.”

“Now that we're in our 60's, I see them worry about what will happen if one or either of them both pass on.”

NEED FOR RESEARCH, EDUCATION, AND AWARENESS

“I think that there are not enough people out there that know about Fragile X. And, it's getting better, but I think more information is needed, and it needs to be brought to the public spotlight. Maybe like autism, I think autism got so much coverage in the past few years, and Fragile X is just as important.”

“My sister and my niece have just done a ton of work with caring for my nephew, but also advocacy work with legislators, family support groups, all kinds of stuff. When they first got the diagnosis with their son, there was really not a lot of these support services available. A lot of physicians did not even consider that diagnosis when children were having certain problems.”

“I hope the study helps you guys. I know it's important, and, we would do anything for my niece and nephew. And for my sister as well. I think this was really important to her”.

“I wanted to participate in this because raising a child with Fragile X and living with Fragile X even beyond adult age is tough on any family member.”

“What I hope to see from this study is more information . . . We think the more information we have to go in our medical file, the better off we'll all be. . . and, that's why the stigma has got to change. You know, we've got to open up and not be ashamed and not be so inhibited in medical research. Thank you so much for giving us the opportunity to open our family up.”

“I saw how my sister took up being an advocate for this kind of thing. And that is one of the reasons as to why I decided to participate in this process here. If I can give an hour plus of my time, that's maybe that's something to think about.”

Section IV: Conclusions

Our research on the sibling generation has revealed new information on the connections that premutation mothers of adolescents and adults with FXS have with their own siblings. We see that siblings vary greatly in the type and frequency of contact and in the degree of geographic distance. When considering all kinds of contact together, over half of sibships had contact several times a month or more.

Importantly, siblings also shared with us, in their own words, rich and valuable perspectives on the impact of having a loved one with FXS, including thoughts on family ties, family planning decisions, and considerations for the future. Overwhelmingly, a central theme across these reflections was the love that the siblings have for their sisters and their nieces and nephews with FXS, even in the face of unexpected life challenges.

We are deeply grateful to all the participants in our study. We are eager to continue to learn more about the very unique experiences of all family members of loved ones with FXS. Our goal is to bring awareness to the service and support needs of families, particularly as the sons and daughters with FXS move into midlife and older age.