



Healthy Aging Research Study Newsletter

SC Family Experiences Lab



Lab Director: **Dr. Jessica Klusek**

VOLUME 3

Highlights

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NIH Fragile X
Center of
Excellence

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for Three More Years!

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Saying
Goodbye to
Familiar Faces



A Message to Our Participants:

It has truly been an honor to work with you and your families over the past few years. Whether you've just joined our study, recently completed your third time point, or have been working with us for even longer, your continued support and participation make everything we do possible. Thanks to you, we're able to carry out meaningful research that advances understanding and support for the fragile X community. As we head into another exciting year, we're delighted to share some updates on what we've been working on!



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MAJOR NEWS FOR FRAGILE X RESEARCH!

University of South Carolina Awarded NIH Fragile X Center of Excellence!

The University of South Carolina has received a new **NIH P50 Fragile X Center of Excellence Award** for its project, "*Translation of the FMR1 Premutation Phenotypes Across the Lifespan.*" This Center unites researchers, clinicians, and community partners to advance understanding, early detection, and treatment of fragile X-associated conditions.

Led by Drs. **Jessica Klusek, Jane Roberts, and Abigail Hogan**, the Center's two core projects focus on the premutation by studying: 1) **FXTAS symptom progression in women**, and 2) early social communication indicators in preschoolers.



The effort involves collaborators from *Rush University*, the *Research Foundation for Mental Hygiene*, the *University of Wisconsin–Madison*, the *Medical University of South Carolina*, and the *Donavan Angel Foundation*, with support from [NICHD](#), [NIDCD](#), and [NIMH](#). **This milestone underscores the vital role of the fragile X advocacy community**, especially the National Fragile X Foundation, in turning research into real-world impact.

Enrollment Extended For Three More Years!



Thanks to funding from the NIH P50 Fragile X Center Award, we're excited to share that our study on women with the *FMR1* premutation (ages 35–80) has been extended for three more years!

This means we will be reaching out to ask for your continued participation in the study. The NIH P50 Fragile X Center Award will allow us to continue to follow current study participants for an additional 2-3 years. This marks a major step forward in expanding the impact of this research and your contributions are critical to furthering our understanding of age-related health problems associated with *FMR1* premutation.

We are deeply grateful for your support and contributions to this research, and we hope that you will continue to be part of this meaningful work!



Recruitment Update – Thank You for Your Support!

The team completed over **100 assessments** this year! A huge thank you to our project coordinator, **Blythe Vickery**, who has been the primary point of contact and support for participating families, **and to all of the women who have taken part - this work would not be possible without you!**



Dr. Jenner - The First LivJoy Postdoctoral Fellow

The [LivJoy Foundation](#) has partnered with the University of South Carolina to fund a fellowship dedicated to **advancing research that improves outcomes for females with FXS!**



Dr. Jenner, the current LivJoy Fellow, earned her Ph.D. in psychology in the United Kingdom, where she focused on autism in genetic syndromes. She is now mentored by a multidisciplinary team of FXS experts at University of South Carolina: **Drs. Jessica Klusek, Jane Roberts, and Abigail Hogan**. Together, they are working on new studies that address the long-overlooked area of female-focused research in FXS.

The LivJoy Fellowship demonstrates how targeted funding, collaboration, and advocacy **grounded in lived experience** can drive progress in closing long-standing gaps in research and care – we're deeply grateful for their support. *Learn more about LivJoy [here](#).*

New Research on Mothers with the Full Mutation!

Dr. Jessica Klusek and Dr. Lauren Jenner have been awarded a \$40,000 pilot grant from the [Carolina Autism and Neurodevelopment \(CAN\) Research Center](#) for a pilot study focusing on mothers with the **fragile X full mutation** (>200 CGG repeats), aged 35-80 years.



While most prior research has centered on individuals diagnosed with FXS in childhood or on mothers who are premutation carriers, **mothers with the full mutation have largely been overlooked**. This new project aims to explore the healthcare needs of these mothers and understand how their experiences may differ. The study builds on Dr. Lauren Jenner's ongoing work, supported by the **LivJoy Foundation**.

Participation includes a one-day in-person visit so we can learn about your experiences. You'll receive \$100!

Do you know a mother (aged 35-80 years) with the full mutation? Perhaps a daughter, sister, niece or friend? We'd love to hear from you! Please contact Lauren on ljenner@mailbox.sc.edu

STUDENT SPOTLIGHT

We want to congratulate Thomas Christensen, doctoral student in Dr. Jessica Klusek's SC Family Experiences Lab, for being recognized by the [Arnold School of Public Health's "I Am Public Health."](#) for his passion and research supporting individuals with developmental disabilities. He has also been awarded the [Randi J. Hagerman Summer Scholar Research Award](#) from the [National Fragile X Foundation](#) to study how social determinants of health impact symptoms of the *FMR1* premutation in women. We are lucky to have you! Watch Thomas' 15-minute video presentation of his project [here!](#)



Thomas Christensen





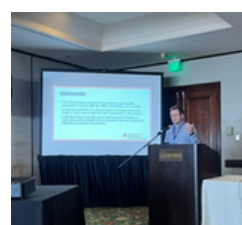
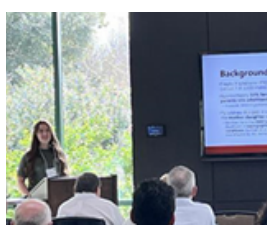
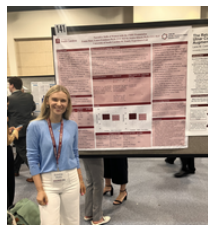
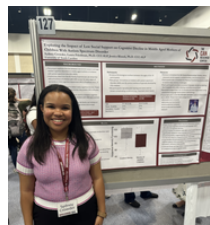
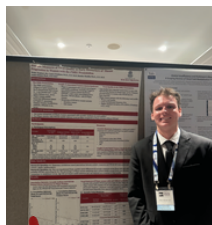
WHAT HAVE WE BEEN UP TO?

Conference Presentations:



Our team has been busy sharing research at national and international conferences this year, highlighting important advances in understanding the *FMR1* premutation and related conditions:

- **Wesley Kephart** (graduate student) presented a poster titled “Acoustic Measures of Vocal Quality as Early Biomarkers of Disease Progression in Women with the *FMR1* Premutation” at the [2025 Fall Voice Conference](#) in Charlotte, NC.
- Undergraduates **Sydney Crowder**, **Emma Hutz**, and **Morgan Rachmiel** presented their research at DiscoverUSC, with Emma earning first place for her poster - congratulations to all three for their outstanding work!
- **Dr. Lauren Jenner** (postdoctoral fellow) presented her study, “The hardest part is allowing her to fall”: Mothers with the *FMR1* Premutation and Their Experiences Raising Daughters with Fragile X Syndrome, at the [2025 Gatlinburg Conference](#) (San Diego) and the [South Carolina Autism and Neurodevelopmental Disorders Consortium \(SCAND\)](#).
- **Dr. Laura Friedman** (former postdoctoral fellow) presented research on sleep, memory, language, and mental health in women with the *FMR1* premutation and mothers of autistic children at multiple conferences (Gatlinburg Conference, [NFXF International](#), [Fragile X International Congress, Barcelona](#)) and received the Best Poster Award at the SCAND Consortium.
- **Thomas Christensen** (doctoral student) presented at major meetings including the [Fragile X Premutation Conference](#) (Italy), Gatlinburg, [ASHA Convention](#), and NFXF International, highlighting work on social determinants of health, language processing, and FXTAS-related motor and cognitive symptoms in women with the *FMR1* premutation.
- **Dr. Jessica Klusek** shared findings at multiple conferences, including Gatlinburg, Fragile X International Congress, NFXF, and the Fragile X Premutation Conference (Italy) on predictors of tremor progression, motor and cognitive outcomes, and mental health in women with the *FMR1* premutation.



Advocacy Events:

Our lab has been proud to take part in key advocacy events at both the state and national levels, joining families, self-advocates, and professionals to raise awareness and support the Fragile X community.

Over the past year, our lab attended **National Fragile X Advocacy Day at the South Carolina Statehouse**. The event featured speakers from USC, the Greenwood Genetics Center, and the Fragile X community who shared stories to raise awareness and inspire change. Dr. Jessica Klusek and Dr. Lauren Jenner also represented the lab at the **21st National Fragile X Foundation Advocacy Day in Washington, D.C.** They joined families, self-advocates, and providers nationwide to share ongoing research and advocate for the fragile X community. We look forward to attending these events again next year and hope to see you there!



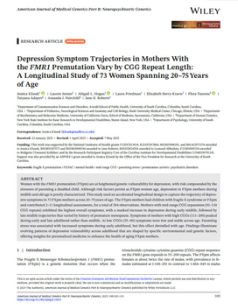
Published Papers:



Depression Symptom Trajectories in Mothers With the *FMR1* Premutation

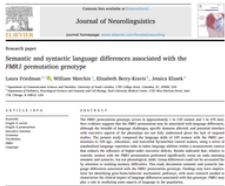
We are pleased to share that **Dr. Jessica Klusek** and **Dr. Lauren Jenner** (postdoctoral fellow) have published a new paper in the American Journal of Medical Genetics Part B: Neuropsychiatric Genetics titled "**Depression Symptom Trajectories in Mothers With the *FMR1* Premutation Vary by CGG Repeat Length: A Longitudinal Study of 73 Women Spanning 20–75 Years of Age**". This study followed 73 mothers with the *FMR1* premutation across adulthood (ages 20–75) to understand how depression symptoms may change across the lifespan. The results showed that experiences are not the same for everyone - they depend on both genetic and life factors:

- Women with **mid-range CGG repeats (91–110)** tended to show the highest depression symptoms, especially around midlife. Women with **lower repeats (55–90)** reported consistently low symptoms across the lifespan, while those with **higher repeats (111–200)** showed more symptoms in early and later adulthood.
- Mothers who experienced **early or premature menopause** were more likely to have **persistent depression symptoms into their 60s**, especially if they also had **mid-range repeats**.
- Raising a child with FXS can increase stress and place additional demands on parents, which elevated depression symptoms.



It is important to emphasize that these findings reflect patterns of risk, not inevitability. Having the *FMR1* premutation and raising a child with FXS does not mean a mother will develop depression, but it may increase the chances of experiencing symptoms. Recognizing the genetic and environmental factors that contribute to this risk can help families and healthcare providers plan for mental health support proactively, rather than reactively. You can read more [here](#).

Semantic & Syntactic Language Differences Associated with the *FMR1* Premutation



Dr. Laura Friedman and **Dr. Jessica Klusek** published a study in the Journal of Neurolinguistics showing that women with the premutation had **more difficulty repeating phrases and sentences**, suggesting subtle language challenges with words and grammar. These differences were **independent of broader cognitive skills**, revealing unique language patterns linked to the premutation - offering new insights into its effects on brain and behavior. You can read more [here](#).

Supporting Self-Determination in Fragile X Syndrome

Dr. Laura Friedman, with collaborators **Dr. Carly Moser** and **Dr. Jessica Klusek**, published a study in the Journal of Intellectual Disability Research examining self-determination - **the ability to set and pursue personal goals** - in young adults with FXS. The study found that while families encourage goal setting, these **opportunities are more limited at school for individuals with greater social communication challenges**. The findings emphasize the importance of fostering goal-setting and decision-making opportunities for all young people with FXS to support independence. You can read more [here](#).



COVID-19 Impacts on Mothers' Mental Health

Former doctoral student **Dr. Carly Moser**, with collaborators **Dr. Laura Friedman** and **Dr. Jessica Klusek**, published a study in Research in Autism Spectrum Disorders on how the COVID-19 pandemic affected mothers of autistic children and children with FXS. The study found that while all mothers faced increased caregiving demands, **those raising children with FXS or autism experienced far greater strain and less social support**. These mothers reported higher anxiety and depression, with many noting the pandemic's direct impact on their mental health. You can read more [here](#).

The effects of the COVID-19 pandemic on mental health in mothers of autistic children and mothers of children with fragile X syndrome

Carly Moser¹, Laura Friedman, Katherine Bergert¹, Alexandra Hickey¹, Jennifer Lee

Research Autism, 10.1016/j.rasd.2020.100000

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Highlights

- Over half of mothers of autistic children had depression/anxiety during the pandemic.
- In a number of children with FXS, anxiety was reported by 42% and depression by 31%.
- Social support was lower for mothers of children with disabilities than controls.
- Social support buffered the decline in psychological well-being due to the pandemic.



By the Way...We Are Still Recruiting!

We are hoping to enrich our recruitment for the following groups of women...



- **Women with the *FMR1* premutation** (55-200 CGG repeats) who are aged **70-80 years** and/or from racial backgrounds that are historically underrepresented in research, such as **Black and Hispanic women**. We are also particularly interested in enrolling more women with **FXTAS**.
- **Women without** a fragile-X associated condition, aged 35-80 years who have biological typically developing children, local to the Columbia, SC area.



Participation involves one to two days of in-person assessments examining language, cognition, and motor skills. Participants will receive up to \$200 in reimbursement. *Please share this opportunity with your family and friends!*

Interested in taking part in our research?

Please contact us on scfamilystudy@gmail.com

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<https://scfamilystudy.com/news/>



Remember, you can also reach our lab by email or phone — you'll find the details at the bottom of the page!



IT'S NOT GOODBYE, JUST SEE YOU LATER!



We are delighted to celebrate the exciting next steps our team members are taking as they embark on new chapters in their academic and professional journeys. While we will certainly miss seeing them in the lab each day, we are incredibly proud of their accomplishments and look forward to staying connected as they continue to grow in their careers.



Dr. Laura Friedman, former postdoctoral fellow, has accepted a position as **Assistant Professor** in the **Department of Speech, Language, and Hearing Sciences** at UT Austin—congratulations, Laura, they are lucky to have you! We are also celebrating our research specialists: **Josh Estrada** is beginning a master's program in **Psychometry** at **Clemson University**, **Christina Koenig** has started the **Genetic Counseling** master's program at the **University of South Carolina**, and **Nhi Ngo** has started the **Genetic Counseling** master's program at **Medical University of South Carolina**.



You may recognize these familiar faces from your visits, and we know you'll join us in celebrating them as they continue spreading knowledge about FXS!



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