Fragile X
Association of Michigan

eXtra, eXtra
Volume XX, Issue 4, October 2024

Mark Your Calendar

Support Meetings

When:
October 5, 2024
February 1, 2025
More info on page 3.

Business Meeting
10:15 - 11 am

Support Meeting
11 am - 12 pm

Where:
Virtual via Zoom
https://us06web.zoom.us/j/5604266326

Details will also be on FXAM FB page and FXAM.org

Special Events:

November 2, 2024
FXAM Moms: Let's Do Lunch!
See page 4.

December 7, 2024
FXAM Holiday Party
See page 4.

Fragile X Association of Michigan
FXAM.org

Mary Beth Langan
313-689-3340
mblangan@hotmail.com

PO Box 1414
Troy, MI 48099-1414

Three Cheers for...

Nicholas - has had a very busy summer. He participated in soccer, poly hockey and baseball. A special trip was taken to Chicago to see the Big Boy train; Nicholas and Dad with the train.

MANY THANKS TO THE LIBERATIS!

Nina Liberati recently let the FXAM board know that she and husband Frank are leaving the board. They've been a major part of FXAM for two and half decades and we are so very grateful for their contributions to our FXAM family.

The young parents of three sons joined the board soon after their son Antonio was diagnosed and have been active in many ways. Nina and Frank and their family and friends were the heart and soul of the many FXAM golf outings over the years which funded many of our scholarships and events. They were an amazing support in so many ways.

Nina says we'll still see them at FXAM events but they need to focus more on their exciting nonprofit Supporting Independence in Michigan. They'll hopefully be breaking ground next year on a home for Antonio and peers. Read about the group at **Supporting Independence in Michigan**. We'll also host them at a future meeting to update us on how the plans are going. We all have more to learn from the Liberatis!

*Don't forget you can also make your day better by dining on their delectables from Liberati's Italian Deli and Bakery **liberatisdeli.com**. The staff is always nice to our son Andrew when we stop by because they know Antonio and so they understand Fragile X! It's so worth the trip!*

2024-2025 FXAM Business and Support Meetings

We've had ups and downs with FXAM meeting attendance since I joined in 2002. Many times due to weather, family obligations and more. At some meetings, especially when we had a speaker, we had great numbers! At some meetings, only a few of us would show up. While even two parents can sometimes have an important discussion during these times, it can be very difficult to make plans to drive to a meeting to have no one show up.

Since Covid, our meetings - both in-person and virtual options - have not been well attended. We've made a different schedule for this 2024-2025 season. Please let us know what you think - now or later, whenever you have comments or ideas, please share! We are open to new thoughts on the subject. We are grateful to be able to share knowledge and experiences with FXAM families - new and seasoned ones - at meetings, at events, in our newsletters and on our Facebook page.

October 5, 2024 was held virtually (7 parents attended)

February 1, 2025 will be virtual only.

10:15 - 11 am Business / 11 am - Noon Support

April 5, 2025 *and* June 7, 2025 will have virtual and in-person options.

Exact time and venue to be determined.

Zoom Link for all virtual meetings:
https://us06web.zoom.us/j/5604266326

Please watch FXAM.org and our FXAM Facebook page for changes and/or confirmation of all meetings and events.

University of Michigan Fragile X Clinic

The University of Michigan Fragile X Clinic is accepting new patients! The clinic manages the care for both children and adults with Fragile X syndrome. Clinicians include a geneticist, genetic counselor, and developmental behavioral pediatrician.

Appointments can be scheduled in the UM Fragile X Clinic by requesting a referral from your child’s primary care physician or another specialist who follows them. More information is available by calling 734-764-0579 or by emailing **UMFragileX@med.umich.edu**

People who are seeking appointments for FXTAS can contact the East Ann Arbor Health and Geriatrics Center at 734-647-5670. Please ask for an appointment with Dr. Peter Todd in the Ataxia Clinic.

> **“Incredible change happens in your life when you decide to take control of what you do have power over instead of craving control over what you don't.”**
>
> *-Steve Maraboli*

What's going on ...

Through the Maze - NPR and Fragile X

Fragile X held him back. An experimental drug is helping him break free - a four-minute listen on NPR (and accompanying story).

npr.org/sections/shots-health-news/2024/09/22/nx-s1-5076913/fragile-x-experimental-drug-helping-autism-adhd-intellectual-disability

The story mentions Zatolmilast which is BPN14770 and part of the Tetra Research Study. The two closest clinics participating in this research are at Rush University Medical Center (Chicago) and Cincinnati Children's Hospital.

See pages **8** and **9** for details on the Tetra Study at Rush.

Fun Fact: Tetra Therapeutics is located in Grand Rapids, Michigan and is now a subsidiary of Shionogi, a pharmaceutical company started in 1878 in Japan.

FXAM Mom's: Let's Do Lunch!

Saturday, November 2, 2024 at 11:30 a.m.

Crispelli's Bakery & Pizzeria
6690 Orchard Lake Rd.
West Bloomfield, MI 48322
248-254-4760
crispellis.com

Let's get together for some Mom-only time!
Email **mblangan@hotmail.com** if you are not on FB.

Please note: This is not a free FXAM event. Each FXAM Mom will be paying for her own food/drinks

Holiday Party - Our Annual Potluck Gathering

Saturday, December 7, 2024 at 5 p.m.

Let's celebrate and look ahead to 2025 with our FXAM Family
at the home of Andrew, Ted and Mary Beth
51 Greenbriar, Grosse Pointe Shores, MI 48236

Some parents make it a date night. Some bring the whole family. Whatever works for you!

Please watch the FXAM FB page in November regarding the # attending
and what delectable you'll add to the gathering.

Contact Mary Beth at **mblangan@hotmail.com** or 313-689-3340.

An Introduction to the Cincinnati Fragile X Center

While at the 19th International Conference, I had the opportunity to meet with some of the individuals associated with the Cincinnati Fragile X Center. Like Rush University they are very active in the world of Fragile X research. The following is what Hannah shared with me about the Cincinnati clinic. Sally

My name is Hannah Sachs. I am a Research Coordinator that works at the Cincinnati Fragile X Center. We appreciate you stopping by our table at the recent NFXF Conference in Orlando! I wanted to reach out to you directly to give you an overview of our clinic and give you opportunities to participate in research or get established clinically.

Our clinic sees a wide range of patients with Fragile X. We see all levels of ages and all levels of functioning as well. We have three primary medical doctors that see our Fragile X patients: Dr. Craig Erickson, Dr. Ernest Pedapati, and Dr. Kelli Dominick. In addition, we have a psychologist team who sees our patient population as well. They focus on the behavioral aspect of care, while the other three look at medication management, behaviors from a pharmacology approach, collaboration with other medical needs, etc. We have clinical psychologists that are able to offer virtual therapy sessions in a variety of states.

Our program also has a strong research component as well. Some people just want to meet with our providers to discuss medications and/or behaviors, questions, concerns, etc. Some people want to do this as well as participate in research studies (we have studies that look at fragile X protein levels, cognitive testing, IQ testing, developmental testing, eye tracking, medication trials, etc.). If you're interested in the research aspect of our Fragile X program, we can get those conversations moving as well. For FX carriers, the options are a little more limited for research, but we can certainly discuss more if interested.

This is a brief overview of the Cincinnati Fragile X Research and Treatment Center. I would be happy to answer any questions you may have via email or hop on a quick call with you to discuss further as well. If you are interested in any of our services or research, we can discuss what the next steps would be to do that because there is additional information I would need to gather from you to move forward.

Hannah Sachs, MPA, *Clinical Research Coordinator III, Behavioral and Developmental Neuropsychiatry*
Division of Child Psychiatry

Cincinnati Children's Hospital Medical Center
3333 Burnett Ave, MLC 4002
Cincinnati, OH 45229
Cell: 317-730-2120 **Desk:** 513-636-2592 **Fax:** 513-803-0072
Fragile X Line: 513-803-1150 **Autism Line:** 513-636-0523

> "Don't expect everyone to understand your journey,
> especially if they've never had to walk your path!
>
> ~ *Anonymous*

Research - Cincinnati Children's Hospital Medical Center

Listed below are studies that we are currently recruiting for. Families are encouraged to reach out to fragilex@cchmc.org if they are interested in participating or have any questions!

1. **Single Dose Drug Challenge Study:** This study is looking to see if medication can treat excessive sensitivity to sounds which is a common and distressing symptom of FXS. Three medications (Baclofen, Memantine, Roflumilast) and a placebo will be tested. Adults, male and female, ages 18-45 years old may be eligible.
2. **FORWARD-March:** This study uses data from families who have a family member with FXS and the clinicians they see to advance our overall understanding of FXS, from the diagnosis, to the evaluation and treatment of it in day-to-day life.
3. **Study of Visual Perception in Males with FXS:** This study aims to learn more about how distracting sensory stimuli impact visual processing and brain activity.
4. **ENTRAIN:** This study aims to improve learning and decrease sensory hypersensitivity in adolescents aged 5-15 with FXS by playing special sounds through headphones to normalize brain activity.
5. **EEG Study of Brain Activity:** This study is looking at how the brain works and changes over time in FXS.
6. **Study of Cognition and Language in Adults with FXS:** This study's goal is to better understand the brain basics of specific cognitive and language impairments in FXS.
7. **Intellectual Disability and ADHD:** A research study to see if executive functioning and processing speed in individuals diagnosed with intellectual disability and ADHD will improve with the study drug, Quillivant.

Research - Resources and Registries

ClinicalTrials.gov - search by condition, i.e., fragile X Syndrome, Angelman Syndrome, Rett Syndrome, etc.

The Fragile X Research Registry - is a database of people who want to be notified about fragile X research studies. It is a confidential, convenient way for families to connect with studies and move research forward—there is no cost or obligation.

FORWARD MARCH - is the next step following a highly successful research study called FORWARD (Fragile X Online Registry With Accessible Research Database) that was started in 2012.

The FORWARD study created the largest database of information on FXS in the United States. Data from the FORWARD study is being used by researchers to learn about the lives of people with FXS. Thanks to families who contribute to FORWARD, researchers are learning about important things like medication use, behaviors, and development over time. These findings are shared with other researchers and clinicians in order to help develop targeted therapies and treatments.

International Fragile X Premutation Registry - The International Fragile X Premutation Registry was created to facilitate and encourage Fragile X premutation research, including future medication and non-medication treatment and intervention studies that could positively impact your quality of life. Fragile X premutation-associated conditions include Fragile X-associated tremor/ataxia syndrome (FXTAS) and Fragile X-associated primary ovarian insufficiency (FXPOI).

Our Fragile X World- **Registry is open, signup to learn about studies!** Our Fragile X World is a research community dedicated to gathering practical information about the experiences of individuals with fragile X syndrome and their families. We conduct studies and share findings in order to advance science, improve professional practice, and impact policy.

Research - Northwestern University

Study Title: A Family Genetic Study of Fragile X Syndrome and FRM1 Premutation

Principal Investigator: Molly Losh, PhD

The Neurodevelopmental Disabilities Lab at Northwestern University is extending successful work conducted through the Family Genetic Study of Fragile X Syndrome with new funding we were awarded from the National Institutes of Health. In this new phase of the project, we are working with women who carry the FMR1 premutation and individuals with fragile X syndrome to study the brain and gene basis of language and related skills in individuals with the FMR1 premutation and their family members. In particular, we are interested in understanding how you use language during different language interactions, how looking and speaking are related, and how the brain processes language by looking at neural responses to speech sounds.

Participation involves completing different language tasks, solving puzzles, looking at screens, completing noninvasive measures of brain activity, and providing a blood sample. Participation can be completed during 1 or 2 visits on Northwestern University’ Evanston campus or at a location that is more convenient for you, and you will be compensated for your time as well as any costs related to your travel. Compensation varies by the amount of tasks you complete.

If you would like to participate, email us at:

kylie.robinshaw@northwestern.edu or **familystudy@northwestern.edu**.

You may also call the lab phone at **847-467-2516** to speak to one of our lab members. We look forward to hearing from you!

Research - Rush University Medical Center

RECONNECT Study

Dr. Elizabeth Berry-Kravis at Rush is currently recruiting for a Fragile X syndrome clinical trial, RECONNECT, evaluating an investigational topical treatment that may help FXS-related behavioral issues.

The trial is recruiting:

- Male and female participants with full mutation FXS
- Between the ages of 3 and 29 years of age.

The trial includes 8 total visits, over approximately 21 weeks, with 4 clinic visits and 4 virtual visits.

Please contact the Rush RECONNECT Study Coordinator Anniek van Aarem for more information at:
312-563-7284 or **anniek_m_vanaarem@rush.edu** or visit **fragilexhelp.com**

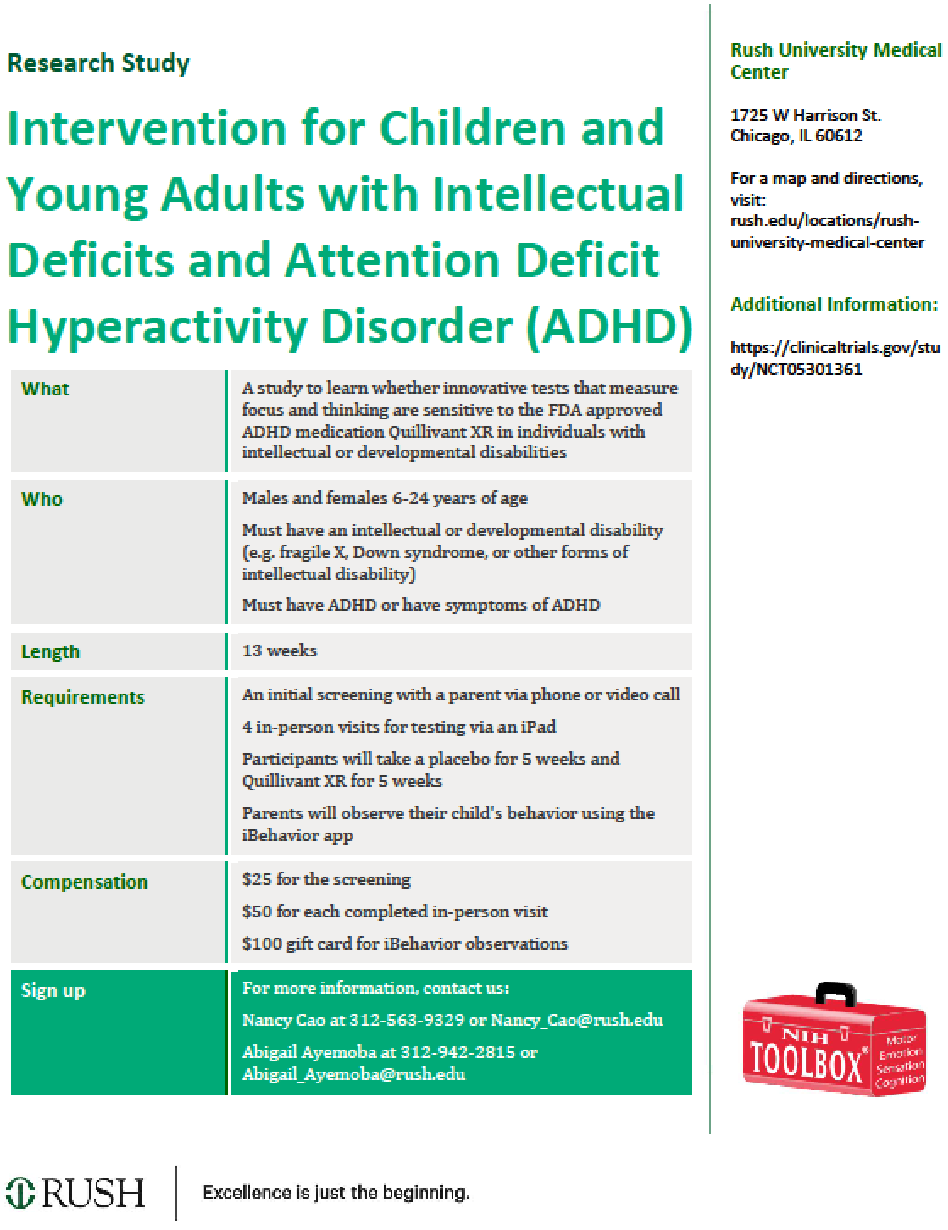
> The business of biomedical research is mostly about failure.
> Few projects we commission will ultimately result in success.
> But every study we do contributes to the body of knowledge that brings science and society closer to a solution.
>
> ~Kenneth Frazier

Research - Rush University Medical Center

Rush University, contact Nancy_Cao@rush.edu

Toolbox Quillivant study - We are currently looking for participants to enroll in the Toolbox Quillivant Study. We are **especially** looking for **girls and women** and really need participants. The overall Toolbox study seeks to validate the iPad-based cognition test, the NIH Toolbox Cognitive Battery, in populations with intellectual and developmental disabilities. The Quillivant component of the study seeks to measure the sensitivity of the Toolbox to interventions (the FDA approved ADHD medication Quillivant XR). We are seeking individuals ages 6-24 years who exhibit hyperactive and inattentive symptoms and who have an intellectual disability.

There are 4 in-person visits over the course of 13 weeks.



Research Study

Intervention for Children and Young Adults with Intellectual Deficits and Attention Deficit Hyperactivity Disorder (ADHD)

What	A study to learn whether innovative tests that measure focus and thinking are sensitive to the FDA approved ADHD medication Quillivant XR in individuals with intellectual or developmental disabilities
Who	Males and females 6-24 years of age Must have an intellectual or developmental disability (e.g. fragile X, Down syndrome, or other forms of intellectual disability) Must have ADHD or have symptoms of ADHD
Length	13 weeks
Requirements	An initial screening with a parent via phone or video call 4 in-person visits for testing via an iPad Participants will take a placebo for 5 weeks and Quillivant XR for 5 weeks Parents will observe their child's behavior using the iBehavior app
Compensation	$25 for the screening $50 for each completed in-person visit $100 gift card for iBehavior observations
Sign up	For more information, contact us: Nancy Cao at 312-563-9329 or Nancy_Cao@rush.edu Abigail Ayemoba at 312-942-2815 or Abigail_Ayemoba@rush.edu

Rush University Medical Center

1725 W Harrison St.
Chicago, IL 60612

For a map and directions, visit:
rush.edu/locations/rush-university-medical-center

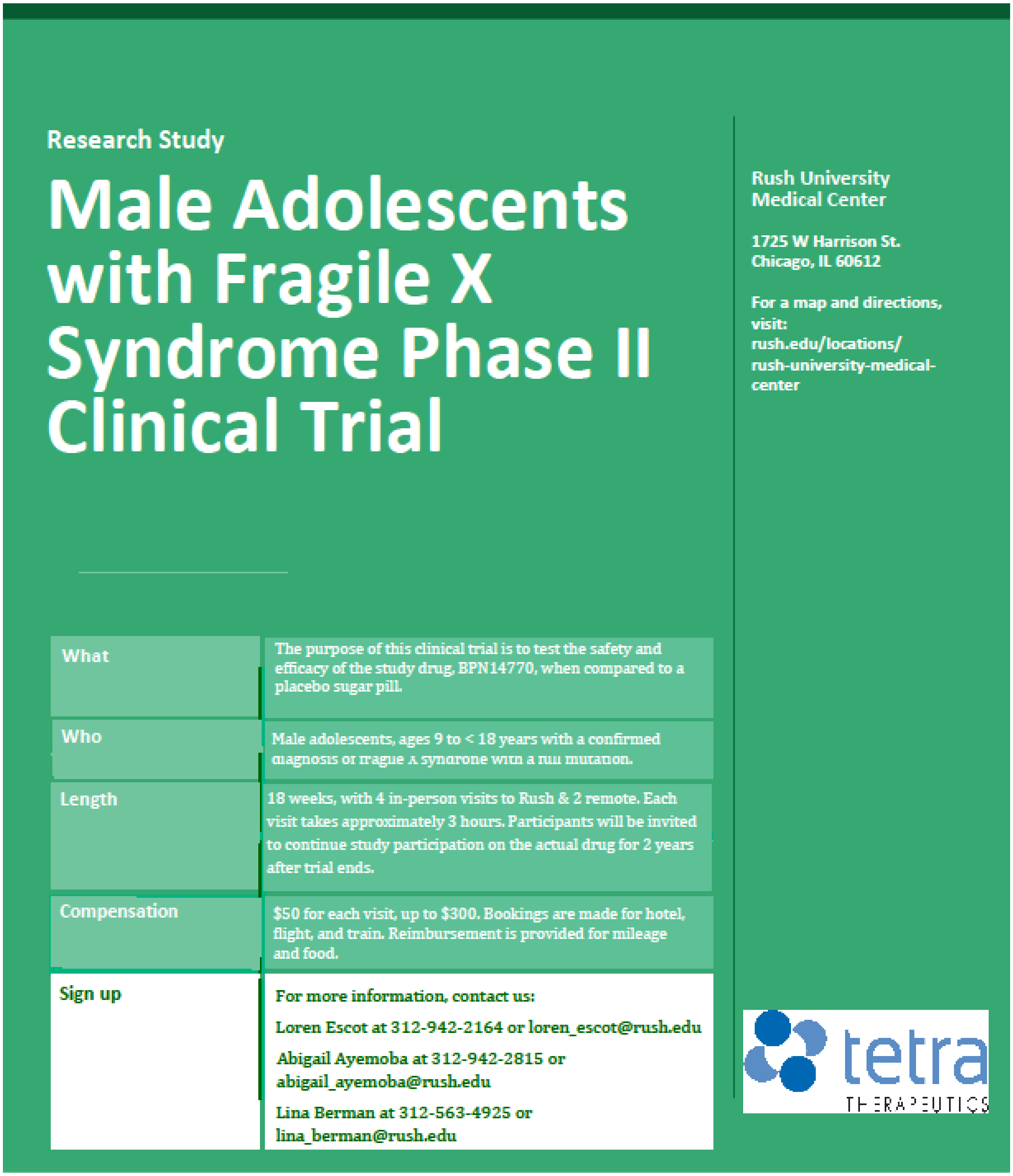
Additional Information:

https://clinicaltrials.gov/study/NCT05301361

RUSH | Excellence is just the beginning.

Research - Rush University Medical Center

For details on the Rush Tetra Research, contact Lina_Berman@rush.edu



Research Study

Male Adolescents with Fragile X Syndrome Phase II Clinical Trial

Rush University
Medical Center

1725 W Harrison St.
Chicago, IL 60612

For a map and directions, visit:
rush.edu/locations/rush-university-medical-center

What	The purpose of this clinical trial is to test the safety and efficacy of the study drug, BPN14770, when compared to a placebo sugar pill.
Who	Male adolescents, ages 9 to < 18 years with a confirmed diagnosis of fragile X syndrome with a full mutation.
Length	18 weeks, with 4 in-person visits to Rush & 2 remote. Each visit takes approximately 3 hours. Participants will be invited to continue study participation on the actual drug for 2 years after trial ends.
Compensation	$50 for each visit, up to $300. Bookings are made for hotel, flight, and train. Reimbursement is provided for mileage and food.
Sign up	For more information, contact us: Loren Escot at 312-942-2164 or loren_escot@rush.edu Abigail Ayemoba at 312-942-2815 or abigail_ayemoba@rush.edu Lina Berman at 312-563-4925 or lina_berman@rush.edu

tetra THERAPEUTICS

RUSH | Excellence is just the beginning.

Research - Rush University Medical Center

For details on the Rush Tetra Research, contact Lina_Berman@rush.edu

Research Study

Male Adults with Fragile X Syndrome Phase III Clinical Trial

Rush University Medical Center

1725 W Harrison St.
Chicago, IL 60612

For a map and directions, visit:
rush.edu/locations/rush-university-medical-center

What	The purpose of this clinical trial is to test the safety and efficacy of the study drug, BPN14770, when compared to a placebo sugar pill.
Who	Male adults, ages 18 to < 45 years with a confirmed diagnosis of fragile X syndrome with full mutation.
Length	18 weeks, with 4 in-person visit to Rush and 2 remote. Each visit takes approximately 3 hours. Participants will be invited to continue study participation on the actual drug for 2 years after trial ends.
Compensation	$50 for each visit, up to $300. Bookings are made for hotel, flights, and train. Reimbursement is provided for mileage and food.
Sign up	For more information, contact us: Loren Escot at 312-942-2164 or loren_escot@rush.edu Abigail Ayemoba at 312-942-2815 or abigail_ayemoba@rush.edu Lina Berman at 312-563-4925 or lina_berman@rush.edu

tetra
THERAPEUTICS

RUSH | Excellence is just the beginning.

Research - Rush University Medical Center

For details contact Emily_C_Timm@rush.edu

ORA: 20021404-IRB01 Date IRB Approved: 4/11/2020 Amendment Date: 4/29/2023

Research Study

FXTAS Research Opportunity

What	**We are researching brain activation during cognitive and walking assessments in people with FXTAS using wearable functional near infrared spectroscopy (fNIRS) technology.** You will be asked to: – Have your walking tested while wearing multiple sensors on the outside of your clothing and a brain imaging cap – Perform a series of thinking tasks and functional balance tests – Have a short neurological examination – Receive an MRI within two months of your initial research visit
Who	Men and women with a clinical diagnosis of FXTAS
Length	Testing will last approximately 2 hours + 1 hour MRI and can be completed in one visit or split into two
Requirements	**Participants must:** – Be ≥ 50 years of age – Have the ability to: 1) Balance and ambulate unaided for 5 minutes, and 2) Follow step-by-step directions **Participants must NOT:** – Have had any lower limb orthopedic surgeries within the past 12 months – Have any other neurological/musculoskeletal disorders that impact mobility – Have a history of significant head trauma
Compensation	Individuals will receive a $100 gift card for participation in this study
Sign up	Email Emily Timm directly at Emily_C_Timm@rush.edu

RUSH | Excellence is just the beginning.

Rush University Medical Center

Armour Academic Facility
Suite 505
600 S. Paulina Street
Chicago, IL 60612

Jelke Building
Suite 871
1750 W. Harrison Street
Chicago, IL 60612

Kellogg Building
Suite 235
1620 W. Congress Parkway
Chicago, IL 60612

Joan & Paul Rubschlager
Building
Suite 5156
1520 W. Harrison Street
Chicago, IL 60607

Research

FXS Caregivers

Help Advance Gene Therapy Research

for Fragile X Syndrome (FXS)

Gene therapy holds promise for FXS treatment, but there's a need for more information on caregiver perspectives.

By completing our anonymous survey, you will contribute valuable insights on your knowledge, attitudes, and perspectives regarding gene therapy for FXS. Your input will help:

- Advance Gene Therapy Research: Provide crucial data to further scientific studies.
- Inform Clinical Trials: Influence the design and implementation of future clinical trials for FXS gene therapy.

Survey link: https://redcap.link/FXSgenetherapy.survey

Contact: vivian.chen@icahn.mssm.edu

Icahn School of Medicine at Mount Sinai

The 19th International Fragile X Conference

Sally's Briefs

I did send out, via email, some very detailed notes on some of the sessions I attended. If you didn't get them that means you are not in my email group. If you want them, let me know at sallyn423@gmail.com.

Medications for FXS:

- Start **Low**, Go **Slow** - this can alleviate some of the side effects.
- Never Ever start two meds at once or change two meds at once, take it one at a time, unless of course you are in a crisis
- Therapeutic decisions are based on the largest problem symptoms and can be trial and error to find the right meds
- Medication is not necessarily "**when all else fails**"
- Medications can create improvement, but not normalize behavior
- Behavior therapy is helpful, Cincinnati Fragile X offers TeleHealth, and it's not unique to Ohio. Many Drs are now licensed to practice in multiple states
- Treatment should be focused on symptoms, and frequency of the symptoms. If for example, aggression only happens once a year it may be better treated through behavioral strategies and not meds

Panel Discussion Treatment and Interventions

- **Using a visual timer:** Start with counting down practices, i.e. 10, 9, 8, …. Be forewarned, visual timers can create anxiety
- **Dental Appointments**
  - Baby Steps! Start with only a visit, getting a toothbrush and a prize if they offer one
  - Social Stories
  - Have them go with you for a cleaning, be a role model
  - Sometimes a single dose of something like a Xanax may help. **ALWAYS TRY A MEDICATION LIKE THIS BEFORE YOU NEED TO USE IT.** Sometimes, some individuals have an adverse reaction, an opposite reaction to what the med should do.

Females and Fragile X Syndrome

Females have unique challenges

- Don't normalize behaviors
- Don't compare males to females
- Don't underestimate their symptoms
- Don't dismiss things because they are considered mild compared to their male counterparts
- Recognize that **avoidance** is a product of anxiety
- Do see Inside Out 2

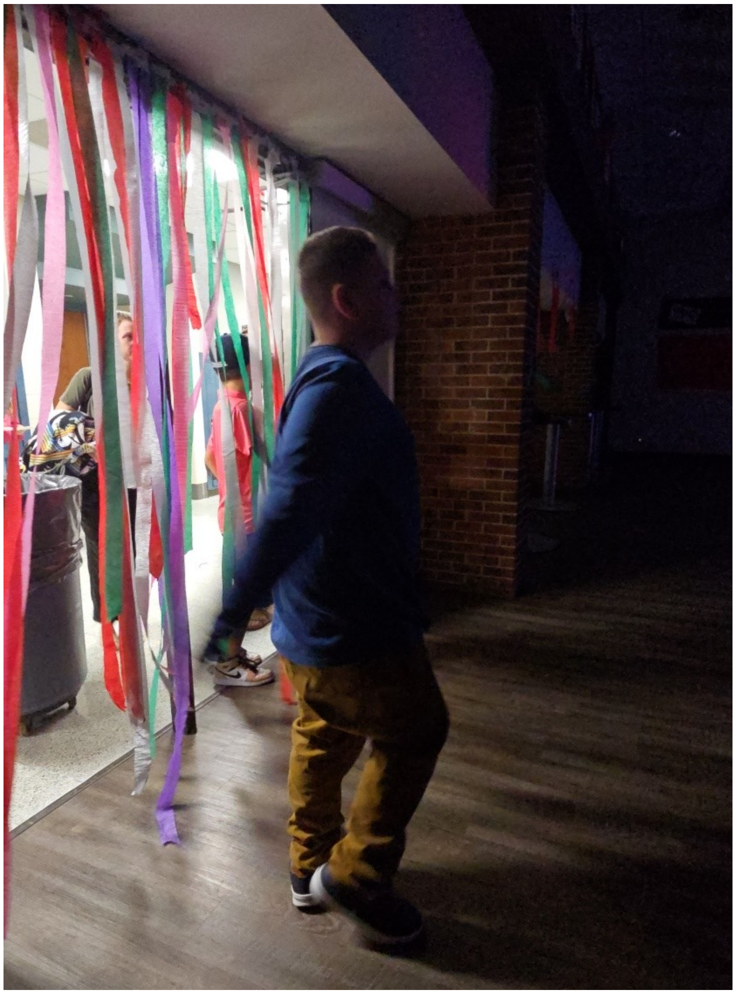
I am thankful I was able to attend the conference, for many this may not be possible. There is a wealth of information available in webinar format at the National Fragile X Foundation (NFXF) and FRAXA Research Foundation (FRAXA) websites.

fragilex.org/family-resources/fxs-premutation-webinars/

fraxa.org/?s=webinar

Three Cheers for Back-to-School…

Cade - went back to school on August 28 and later he attended his first dance. Way to go Cade!



"Autumn leaves don't fall, they fly. They take their time and wander on this their only chance to soar."

~ Delia Owens

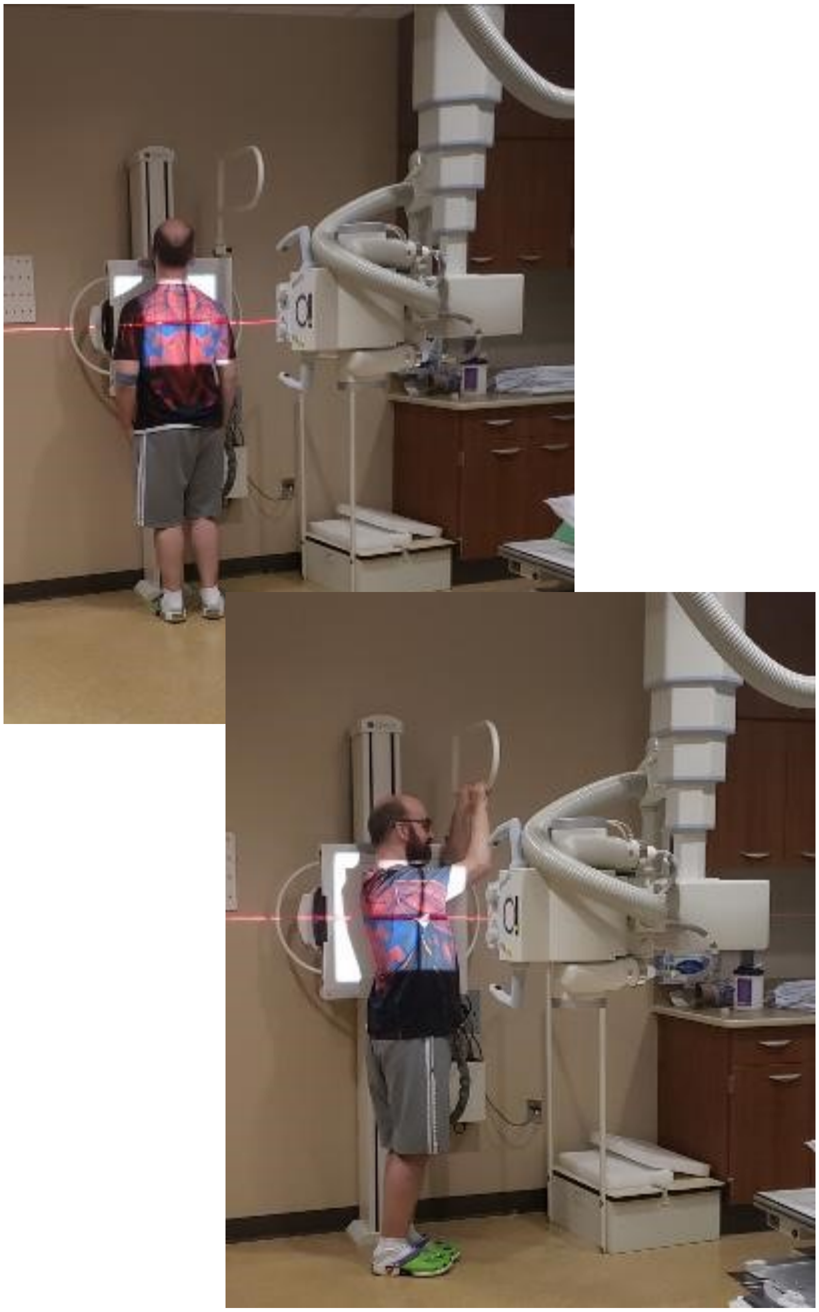
Three Cheers for...

Molly - it's senior year and Molly is sharing one of senior year pictures. Here's to a great senior year Molly!

Annabelle - started horseback riding and she is loving it. She started Eton Academy and is really thriving! Grandpa is still under Dr. Todd's care but is having a hard time finding medications that help with the tremor.

Three Cheers for...

Austin – As we prepare for our snowbird time in Florida, he's had a lot of appointments. He rocked everything! The X-rays were a new adventure. Between his psych, primary doctor and the Northwestern Research Study, he's filled 8 vials for blood work (please note the blood work for Northwestern is optional.)
And by the way, I do let him dress himself.



Fall and the Holidays, a recipe for success by Sally

Have I ever told you how much I used to hate Fall?

Before we kick off the season we experience back-to-school challenges, only to be catapulted into changing weather, days getting shorter, and the "Holiday season”, Halloween, Thanksgiving, Hanukkah and/or Christmas. Oh the anxiety, not only for Austin who has fragile X, but for his mom who would struggle with anxiety over his anxiety!

Fall used to always be rough. At one point, I would actually increase one of Austin’s medications just for fall and decrease it later in January. Whatever helped us through the season was my motto. It doesn’t help that we can walk into a few stores in September and be assaulted not only by Halloween decorations but Christmas as well.

For Austin, we keep a calendar and every morning the first thing Austin does is cross off a day, the second thing he does is ask “What’s for dinner?” Important dates are simply marked with a single, bold, capital letter, i.e., H is for Halloween, T is for Thanksgiving, A is for Austin’s birthday. This keeps him grounded, it’s easier to picture how far away a holiday may be. I will admit schedules can be tricky. For Austin, I’ve never used a daily schedule because it often created anxiety and for us “anxiety” is public enemy number one.

Social Stories (written or video) may also be helpful. We have a list of holiday social stories at fxam.org/resources.html.

You may want to use one of these, or simply use one of these as a template to create your own social story, centered around your family traditions.

Then there is “Plan B”, during the actual holiday. Austin’s dad comes from a very large family, a family gathering could easily have 70 people (adults and children). We always had a “Plan B”, we travelled in two vehicles, Austin knew what event (i.e., desserts or presents) would signify the end. Travelling in two vehicles if I needed to bail early with Austin I could do so without it impacting the entire family. I only had to do this once, when Austin was little. An Uncle who looked like a member of ZZ Top got up close and personal, making strange noises. It pushed Austin over the edge, we were only twenty minutes into the event but I knew I had to get him out of there or it would only get worse. I think he’s done pretty well considering we only had to bail once really early and Austin is now 32.

So take a deep breath and prepare, you can make it easier for your child, with a little planning.

Editors:
Mary Beth Langan 313-689-3340
mblangan@hotmail.com
Sally Nantais 734-486-5636
sallyn423@gmail.com

This newsletter is published quarterly and sent to all members and supporters of FXAM. Permission is granted to reproduce and distribute this newsletter for noncommercial purposes.

Electronic copies of our newsletter are available on our website. If you would like to share them with family, friends or professionals, please share our web address with them.

313-689-3340

mblangan@hotmail.com

FXAM.org