

# Fragile X Research and Treatment Center Newsletter

February 2022



## Meet The Team



Dr. Ernie Pedapati, MD, is a psychiatrist and pediatrician who provides clinical care for individuals with Fragile X of all ages. His research interests include developing new treatments and understanding brain activity. He co-directs the Fragile X gene therapy program at Cincinnati Children's.



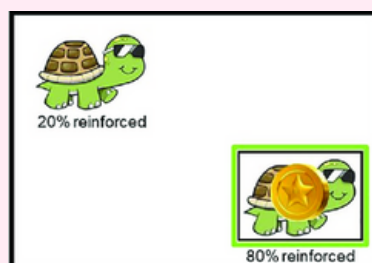
Dakota Gallimore graduated from Eastern Kentucky University with a Master of Arts in Clinical Mental Health Counseling and a Bachelor of Science in Psychology. He is the lead coordinator for the Tetra study and sponsor-initiated trials involving autism spectrum disorder and Fragile X Syndrome.

## Fragile X Results Spotlight

### Parallel learning and cognitive flexibility impairments between Fmr1 knockout mice and individuals with FXS

The probabilistic learning task (shown below) helps us learn about cognitive flexibility, which is the ability to change our behavior in response to changes in our environment. It's been used in humans and rodents to teach us how cognitive flexibility is affected in neurodevelopmental disorders, and how treatments (behavioral and/or pharmacological) can influence cognitive flexibility.

#### ACQUISITION



- Choose correct stimulus
- Immediate feedback ("X" or coin)

#### REVERSAL



- Correct stimulus changed without warning
- Choose new correct stimulus

The participant chooses between animals that are displayed in two locations on the screen. One of the animals is in the "correct" location, and choosing this animal will usually be rewarded with positive feedback (the coin). Like in the real world, the "correct" location is not always rewarded, so participants have to learn which location has the greatest probability of being rewarded by using trial and error. Over time, the participant learns to continue choosing the animal in the "correct" location to get more coins.

After the participant learns which location is usually rewarded, the "correct" location changes (reversal) and the participant has to choose the animal in the new "correct" location to continue receiving coins.

	Males (FXS and mice)	Females (FXS and mice)
<b>Learning Deficit</b>	Had deficits in both acquisition and reversal phases	Had deficits only in the reversal phase
<b>Increased Errors</b>	Related to broader deficits in executive function	Related to more severe psychiatric features (irritability, depression, OCD)

Due to consistent results in both humans and mouse models, this can be used to understand cognitive flexibility and to evaluate the effectiveness of treatments.



Find the full article here:  
<https://doi.org/10.3389/fnbeh.2022.1074682>



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## **Additional Study Opportunities**

**EEG Study of Brain Activity:** This study is looking at the mechanism and brain circuits underlying Fragile X Syndrome. It is a correlational study that enrolls individuals with FXS, their family members and control groups.

**New Parent-Report Measure of Inflexibility in FXS:** Behavioral inflexibility is common in FXS and greatly impacts daily life. We developed a measure to better capture this symptom and need to test it out.

**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.

**FORWARD March:** This study uses data from families who have a family member with Fragile X syndrome and the clinicians they see to advance our overall understanding of Fragile X syndrome, from the diagnosis, to the evaluation and treatment of it in day-to-day life.

**Computer Based Cognitive Testing:** The goal of this study is to develop ways to measure cognition, in hopes we can use these in future clinical trials to help us understand which treatments work.

**Hearing, Language, and the Brain:** This study uses a cap that monitors blood flow and electrical activity in the brain, with the goal of finding a link between hearing, brain activity, and language impairment.

**Interested? Email [FragileX@cchmc.org](mailto:FragileX@cchmc.org)**

## **Study Spotlight**

### **Tetra**

#### **Who?**

Males with FXS (ages 12 to 45)

#### **What?**

This study is assessing the efficacy of BPN14770 in the change of cognition and function in males with FXS. The study's goal is to determine the effectiveness against a placebo. This includes 6 clinic visits and 1 phone call over 15 weeks. There is an open label option at completion.

#### **What's Involved?**

NIH-Toolbox, blood draws, Covid-19 test, ECG, cognitive assessments, and parent measures

#### **Contact**

[Dakota.Gallimore@cchmc.org](mailto:Dakota.Gallimore@cchmc.org)

# THANK YOU!

Our research team at Cincinnati Children's Hospital would like to thank you for your participation and interest in our research studies!

# COMMUNITY EVENTS

*FEBRUARY 2023*

## **Sensory Friendly Maple Magic**

February 11th and 18th from 1 - 2:30pm

Learn the history and mystery of maple sugaring in this family friendly program - tap a maple tree, play a maple game, taste real maple sap, and learn how it turns into maple syrup!

Register at <https://explorenature.regfox.com/sensory-friendly-maple-magic>

## **Executive Functioning and Organizational Skills for Individuals with Developmental Disabilities**

February 13, 2023 from 5:30 - 6:30pm

Register at <https://tinyurl.com/executivefunctioningDD>

## **Sensory-Friendly Princess & Frog**

Wednesday, February 22, 2023 at 1:00pm

The Taft Theatre, 317 E 5th Street, Cincinnati, OH 45202

Special performance for individuals with developmental disabilities and their families (not for general public).

Only a limited number of tickets will be sold.

<https://thechildrenstheatre.com/sensory-friendly-princess-frog/>

## **Sensory Saturday at the Cincinnati Art Museum**

Upcoming dates: February 25th

Email [access@cincyart.org](mailto:access@cincyart.org) to join the mailing list

More resources available at [cincinnatiartmuseum.org/access](http://cincinnatiartmuseum.org/access)

## **Ohio Parent to Parent Support Parent Trainings - Virtual**

Every 1st and 4th Thursday

from 12 - 1pm or 6:30 - 7:30pm

Registration is Required!

# COMMUNITY EVENTS

MARCH 2023

## **Table Talk: Infants and Toddlers**

Tuesday, March 7, 2023 at 7:00pm

With Drs. Kelli Dominick and Betsy Smith

## **Managing Anxiety: Helping Kids with Developmental Disabilities Cope & Succeed**

Wednesday, March 8, 2023 from 5 - 7 pm

For caregivers of children ages 8 and older

Registration opens on February 8th at <https://tinyurl.com/DDanxiety>

## **Adaptive Boxing 2023 Try-It**

March 18th, Imhoff School of Boxing in Hamilton, OH

Ages 6 and up

Register at <https://tinyurl.com/adaptiveBoxing>

## **Adaptive Sports Clinic**

Thursdays from 8am - 12pm

Medical Office Building, 3430 Burnet Avenue

For individuals with disabilities ages 5 and older who need help identifying potential opportunities for sport or recreation, require ongoing physical therapy intervention to safely return to sport, and need to master basic skills in preparation for starting a new sport

Email [otpt.be.well@cchmc.org](mailto:otpt.be.well@cchmc.org) to schedule

## **Adaptive Gaming Night**

Last Wednesday of Every Month (by appointment)

Anyone with a disability interested in gaming who has difficulty playing with a standard Xbox or Nintendo controller

Email [gamers@cchmc.org](mailto:gamers@cchmc.org) to schedule

# UPCOMING EVENTS

## **Adaptive Hiking Series**

Saturdays, April 15th - May 20th

Registration will open soon at:

<https://campscui.active.com/orgs/CincinnatiChildrensHospitalWellness>

## **Cincinnati Fragile X Center Conference**

Saturday, July 22, 2023, 8 am - 5 pm

Cincinnati Children's Hospital Medical Center

# ONLINE PRESENTATIONS

Available at <https://fragilex.org/blog/2022-conference-sessions>

## **Emotion Dysregulation in Fragile X Syndrome**

**Rebecca Shaffer, PsyD**

This presentation focuses on the signs of emotion dysregulation in Fragile X syndrome and ways to respond including practical strategies and an Emotion Regulation Toolkit to take home.

## **Common Behaviors and Available Treatments**

**Across the Lifespan**

**Debra Reisinger, PhD**

This presentation highlights how to recognize behavioral challenges in FXS and the most common behavioral difficulties and treatment options at each developmental stage. Specific tips will be shared that families can easily implement with their children or adolescents.