

THE FRAGILE X RESEARCH REGISTRY

A National Collaboration to Advance Fragile X Research

REGISTRY NEWS 2018

STUDIES COMING SOON

NEW LANGUAGE STUDY

Dr. Audra Sterling, a University of Wisconsin communication researcher, received an NIH award for a new study called “The Relationship between Language and the Brain in Neurodevelopmental Disorders”. The new project will extend her previous studies by adding brain measurement using structural MRI to explore the potential mechanisms influencing the language profiles in fragile X, autism, and Down syndrome. Her new study will be for school aged participants 9-18 years of age.



NEW CLINICAL TRIAL

Dr. Laura Politte will be directing a FXS clinical trial for cannabidiol at the University of North Carolina at Chapel Hill with Zynerba Pharmaceuticals. Dr. Politte provides specialized care for children and adults with developmental, behavioral and psychiatric conditions and conducts clinical treatment studies. She is currently conducting an treatment study to evaluate the benefits of broccoli extract in autism.



For information on studies visit

www.fragileXregistry.org/studies

CURRENT STUDIES

ON-LINE SURVEY ABOUT CHILD BEHAVIOR

Dr. Clare Harrop at UNC-CH and Dr. Brian Boyd at University of Kansas are conducting an on-line survey for parents of children 3-17 with fragile X syndrome. Dr. Brian Boyd and his team are developing the Repetitive and Inflexible Behavior Inventory (RIBI), a parent report measure designed to detect incremental changes in the behavior of individuals with intellectual and developmental disabilities. It can be used to determine the efficacy of interventions targeting behavioral inflexibility and rigidity. These symptoms are characteristic of autism and other neurodevelopmental disorder and often interfere with individual's daily routine and participation in school and activities.



Fragilexregistry.org/studies

PET IMAGING STUDY AT NIMH

Dr. Bebe Smith at the National Institutes of Mental Health is enrolling young men with fragile X 18-24 years of age for an imaging study that uses positron emission tomography (PET) technology. Their goal is to understand how protein formation in the brain is affected in FXS. Participants are invited to the NIH campus in Bethesda, MD near Washington, DC and stay in special accommodations on the NIH campus. Contact Inna Loutaev, 301 496 4707 or E-mail loutaev.inna@nih.gov



The Fragile X
Research Registry
www.fragilexregistry.org

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STUDY RESULTS AND FINDINGS

The FXS Registry has had the opportunity to assist colleagues in the FX research community with recruitment for the following studies. We are pleased to inform you about their progress.

LONGITUDINAL IMAGING STUDY OF INFANTS WITH FXS

Dr. Heather Hazlett, University of North Carolina

Dr. Hazlett and her team have been using magnetic resonance imaging to follow the brain development of infants and toddler with fragile X syndrome and identify potential targets for intervention. In a recent publication, Dr. Meghan Swanson, a post doctoral fellow in the UNC Imaging lab, describes differences noted in the white matter of infants with FXS. Read about their work in the Research Brief, “Brain difference in fragile X are detectable by six months of age.”

<https://www.fragilexregistry.org/Research/Docs/37.pdf>

SKIN CELLS TO STEM CELLS

Dr. Anita Bhattacharyya, University of Wisconsin

Dr. Bhattacharyya and her research team successfully identified a laboratory technique to help detect how genes respond to drug therapies. This technology will a faster and more effective way to screen for drugs that may help turn on the fragile X gene without harming cells. Read about her work in more detail in the Research Brief, “Lighting up the search for a therapy for fragile X syndrome”.

<https://www.fragilexregistry.org/Research/Docs/36.pdf>

ASSESSING PREFERENCES FOR USE OF CLINICAL DATA IN INDIVIDUALS WITH IDD AND THEIR GUARDIANS

Dr. Melissa Raspa, RTI International

Dr. Raspa and her team have surveyed caregivers of individuals with FXS and autism to learn about their preferences regarding the use of their child’s health care data for research purposes. They are exploring a variety of factors which may impact expressed preferences.

Research reports are available at

<https://www.ourfragilexworld.org/reports>

MODIFIERS OF FMR-1– ASSOCIATED DISORDERS

Dr. Emily Allen, Emory University

Dr. Allen, Dr. Stephanie Sherman, and Dr. Steve Warren, veterans in FXS research, have an NIH Fragile X Center award to discover genes that affect the risk and severity of three fragile X-associated disorders: FXTAS, FXPOI, and seizures in males with FXS. Dr. Allen will be presenting on their work at the NFXF Conference in July, 2018.

<https://genetics.emory.edu/fragile-x/research.html>

FAMILY ADAPTATION TO FXS

LANGUAGE DEVELOPMENT OF TEENS AND YOUNG ADULTS WITH FXS

EMERGENCE, STABILITY AND PREDICTORS OF ANXIETY IN FXS

Dr. Jane Roberts, University of South Carolina

Dr. Roberts work focuses on understanding the biological mechanisms that underlie cognitive and behavioral functioning in children and adults with neurodevelopmental disorders such as autism, fragile X syndrome, and AD/HD. Dr. Roberts and her team will be presenting findings at the 2018 NFXF Conference.

A list of publications from Dr. Roberts and her colleagues and links to the articles is available at

<Http://uscdevlab.com/nih-funded-grants/fragile-x-and-autism-research/>

The Fragile X Registry is a unique and valuable resource for the FXS community. Individuals join the Registry on-line; a clinic visit is not required. Members are informed about research studies but are never obligated to participate.