

# Neurodevelopmental Disabilities Lab

NORTHWESTERN UNIVERSITY

## Inside the Research

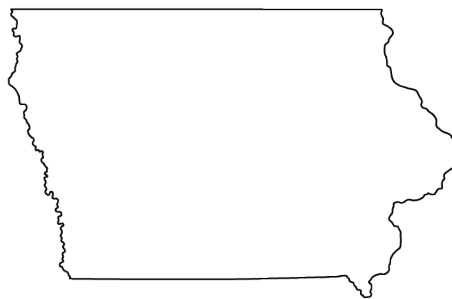
### A Special Thank You!

We are excited to provide an update on the progress of our family studies of autism, fragile X syndrome, and Down syndrome! The time and effort that families devote to research participation is helping us to make important discoveries on a number of fronts, and has resulted in several new publications, new federal funding for our research studies, and new collaborations with networks of professionals and families who are invested in improving support for families. We hope you will consider learning more about our research as we begin to share new study information and participation opportunities with you. We highlight several of these new studies in the pages that follow, and hope you may be interested in participating!



### Find us at the Iowa State Fair

Our group at the NDL will be attending the Iowa State Fair from August 10<sup>th</sup>-August 20<sup>th</sup> in Des Moines, Iowa, and hope we may see you there! We will be recruiting new participants, and also offering opportunities to participate in the study in Des Moines during the fair dates. We hope this may be convenient for families, and also looking forward to the chance to connect and update you on the progress of our work. If you would like to find out about our new studies and think you may want to participate while we are in the area, simply contact our lab before the fair (and if you see us, come say hi!). All families who participate in our studies will receive compensation. We are thrilled to be attending the Iowa State Fair for the first time ever and hope to see you there!



### Overview of Our Research

Our research uses novel methods to study the language and cognitive abilities of individuals with autism, fragile X syndrome, and Down syndrome. Our studies also focus on family members in order to explore how different complex traits run in families, and may relate to genes associated with autism and fragile X. Our studies also examine how the *FMR1* gene may contribute to language and other abilities in carriers of the gene in its premutation state, and in the general population, helping us to understand the genetic basis of complex human traits that we all share.

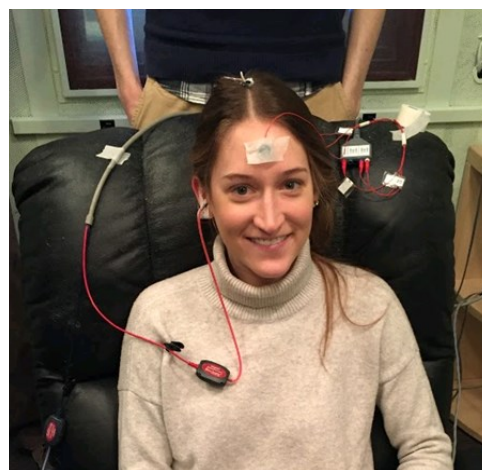
### Contribute to our Understanding of Autism and Fragile X Syndrome!

We are currently looking for families of typically developing children to participate in our research. We would appreciate you sharing our contact information with families in your area who would be willing to contribute their time to help us learn more about fragile X and autism!

## New Studies

### Studies of the brain basis of language

We recently received funding for a second stage of our Family Genetic Study of Autism from the National Institutes of Health. This five-year study will examine how individuals with ASD and their parents represent different sounds of speech in their brains. We do

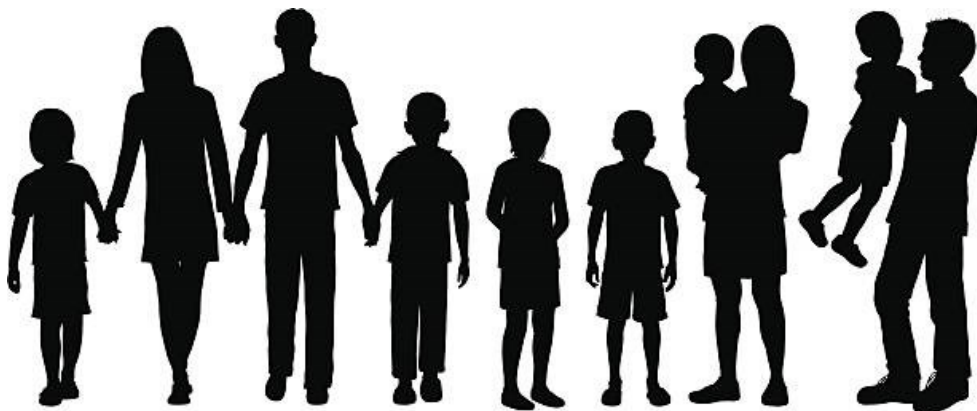


this using the frequency-following response (FFR), which is a measure of sound-evoked brain activity and can tell us about sound processing in the brain. The

FFR provides rich information about how brains process features of sounds, including pitch and timing, and involves listening to different sounds while watching a movie. The information from this task can provide important information about how people process language differently, and whether certain patterns of neural processing of language may be heritable and related to genes involved in ASD.

### New multigenerational studies of autism

Related to our studies of the neural basis of speech, our work also looks at multi-generational predictors of ASD by including great-grandparents, grandparents, parents, and individuals with ASD. In the previous phase of this study, we found evidence that subtle profiles of childhood academic performance in parents, based on



the Iowa Test of Basic Skills (ITBS), predicted features of ASD in their children. This result suggests that different learning patterns during childhood, in people who do not themselves have ASD are linked to the

genes involved in ASD. This is meaningful to the study of ASD in showing how traits related to ASD are genetically based and exist in all of us! We are excited to expand our work to include other family members across generations who went to school and Iowa, and so had taken ITBS in school. We hope this study will help us to better understand how personality, language, and cognitive profiles run in families of people with and without ASD, and which may also relate to unique abilities and their genetic basis.





## Multigenerational studies of fragile X syndrome and women who carry the *FMR1* premutation

Fragile X is a major focus of research in the NDL, where we are also studying the role that the fragile X gene, *FMR1*, plays in language and cognitive development more broadly through studies of individuals who carry the *FMR1* gene in its premutation state. Some important new findings are emerging from our studies of fragile X and the *FMR1* premutation. In particular, through our studies exploring how childhood academic performance on the ITBS in parents is related to their child's symptoms of ASD, we are currently addressing similar questions in families affected by fragile X syndrome (FXS). In particular, we are hoping to better understand how the *FMR1* gene influences personality and academic traits in parents, potentially with interactions of other genes that may also influence clinical symptoms in the next generation of children with FXS. In this new study, we have begun assessing similarities and differences in performance on the ITBS in parents of children with ASD, as well as individuals who carry the *FMR1* premutation. We are starting to see different patterns emerge between these groups. While parents of individuals with ASD show childhood patterns associated with language, females with the *FMR1* premutation show early differences in math. The findings demonstrate how genes that relate to ASD, like *FMR1*, are expressed subtly among people without ASD through language and cognitive profiles. We are also finding that in both parent groups, certain patterns of academic development in childhood predict the presence of ASD-related traits in the next generation of these parents' children. It is possible that these intergenerational patterns, in parents of individuals with two different, yet related, disorders reflect the influence of conducting a number of follow up investigations into these findings, so keep a look out for updated results!



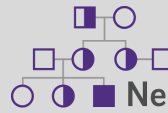
## Visual and sensory processing in autism

We want to thank to all the families who have participated in our study of perceptual skills in individuals with ASD and their relatives! Because of your generosity in devoting your time to participate in this work, we were able to begin analyzing our data ahead of schedule. In our eye tracking studies, we have found that individuals with ASD may view the world differently than those without ASD by using different processing styles that are neither local (detail oriented) *nor* global (seeing a scene as a whole). We are also seeing some subtle similarities in how their parents observe their world, suggesting that visual processing styles might run in families and be linked to genes involved in ASD. We have recently expanded this work to include a non-invasive measure of brain activity (EEG) to explore associations with underlying mechanisms that may run in families.

In addition to our studies of visual processing, we are also exploring other types of sensory processing. Differences in sensory processing, including hyper- and hypo- sensitivity, are a core feature of ASD. Individuals with ASD may also show differences in processing information from multiple senses at the same time, such as integrating sounds and visual stimuli. Multisensory processing is important for language development, including social communication, and the NDL is conducting exciting new research investigating these associations in ASD, and how these features may run in families. This information will help provide clues into the genetic links of autism. We are recruiting participants for these studies currently. Please contact us if you are interested in participating!



**The NDL is on Facebook!**  
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## Upcoming research presentations

We'll be presenting our findings with other researchers and clinicians at two upcoming international conferences. At the International Association for the Scientific Study of Intellectual and Developmental Disabilities (IASIDD), we will share our findings on the longitudinal profiles of social communication (i.e., pragmatic language) in children with fragile X syndrome, Down syndrome, and ASD. These data stem from over 8 years of work following children closely to understand how different language skills develop over the course of childhood. We found that the social communicative abilities in children with fragile X syndrome and ASD showed greater delays than general cognitive ability, and structural language skills like vocabulary and syntax. The presence of autism symptoms in children with fragile X significantly impacted communication development, and a subgroup of children with fragile X showed very similar profiles as those with autism only. These findings may help clinicians to better tailor interventions to specific language skills impacted differently across groups, and also point to the importance of conducting screenings for autism among children with fragile X.

We will also be presenting our data from a study of childhood development of individuals with the *FMR1* premutation at the 3<sup>rd</sup> International Conference on the *FMR1* Premutation. As in our studies of relatives of individuals with ASD, for this study we worked with individuals who carry the *FMR1* premutation and went to school in Iowa and took the Iowa Test of Basic Skills. Our exciting early findings are described above (see *Multigenerational Studies of fragile X*, p. 2), and we're looking forward to discussing the significance and next steps with other leaders in the fragile X research and clinical communities at this focused conference.

**Thank you to all the families and our partners who  
 have supported this important research!**

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