

Neurodevelopmental Disabilities Lab NORTHWESTERN UNIVERSITY

Inside the Research

Letter to our participating families

We are excited to provide you with the latest updates on the progress of our family studies of autism, fragile X syndrome, and Down syndrome. First, we want to thank all of the participating families for their time and effort devoted to this research – you are helping us to make important discoveries on a number of fronts, and we can't thank you enough. 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. Please also consider sharing our information with other families who may be interested in this research. We are actively recruiting for several studies and appreciate any help spreading word of this work.



The NDL at the Iowa State Fair

The NDL is excited to share that we will have a booth at the Iowa State Fair in Des Moines, Iowa! You can find us at the Iowa State Fair in the East side of the Grandstand building (also known as the Halls of Law and Flame), located near the Iowa State Patrol and Iowa Special Olympics. 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 look forward to connect and update you on the progress of our work. We hope to see you there!

Inside the Edition



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Latest multigenerational study reveals unique academic profile in relatives of individuals with ASD

You might remember from newsletters that our earlier studies have found a unique pattern of developmental skills that seem to run in families of individuals with autism, and so could be tied to genes involved in autism. For example, we found that parents of individuals with ASD had more variable development across academic areas, with greater differences in language performance in particular, and which related to their child's symptoms of autism. Working with our collaborators who study large population trends involving data from standardized tests conducted state-wide, our early look at new population data suggests similar unique patterns among individuals later diagnosed with ASD and their siblings. We also have been excited to team up with Project Talent, the largest and most comprehensive study of high school students nation-wide. Many of you may have remembered participating in this study in the 1960s, and we are so grateful to those of you that have participated in our study with Project Talent data. *Continued on p.2*

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.

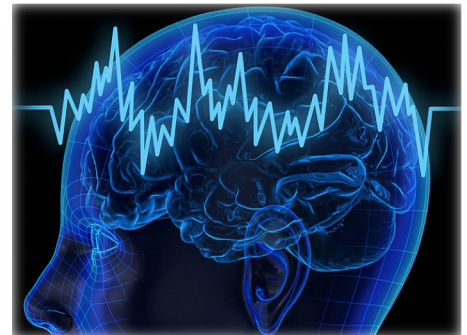


Latest multigenerational study reveals unique academic profile in relatives of individuals with ASD— Cont. from p. 1

Based on this new and exciting work, which was recently presented at the Annual Meeting of the International Society for Autism Research, we again found a unique pattern of development in relatives of individuals with ASD, where we observed a split between reading versus math skills across multiple generations. In parents, a significant discrepancy between reading and math abilities in school was associated with variation in their child's autism symptoms later in life. A split between reading and math skills was also observed among some siblings of individuals with autism, though this did not strongly predict ASD sibling status across the board. Similar reading and math splits were observed in a small sample of grandparents and great-grandparents of individuals with ASD in the Project Talent study. Together, these findings suggest a unique and variable cognitive profile evident across multiple generations (siblings, parents, and grandparents, great-grandparents of individuals with ASD), which may be related to autism-risk genes that are passed across multiple generations within a family. This work could help us to connect the dots between patterns of behavior that can help us better understand the genetics behind autism.

Understanding the Brain and Gene Basis of the Melody of Speech in ASD

Shivani Patel, a doctoral student in the NDL, has led new research in the NDL to study the brain and gene basis of speech prosody (i.e., the melody of your speech) differences in ASD. In a study presented at the meeting for the International Society for Autism Research, Shivani reported differences she found in the way individuals with ASD and their parents use what they hear themselves say to adjust their voice pitch. This is something we all do without realizing it, such as adjusting our pitch when we hear our voice crack. In this study, participants said “ahh” or asked a question and heard themselves doing so through headphones. While participants were vocalizing and listening to themselves speak, we randomly made the pitch of what they were listening to higher or lower to see how they used what they heard to modify what they were saying in the moment. We found that individuals with ASD exhibited larger changes in voice pitch in response to the pitch changes in what they heard while saying “ahh” and asking a question. Additionally, brain responses measured during this task suggested neural differences in the detection of the pitch changes in individuals with ASD. Parents of individuals with ASD also demonstrated larger responses to pitch changes in what they heard while saying “ahh” but varying response patterns when asking a question, indicating different mechanisms of integrating of what was heard and what was being said. Importantly, larger responses to pitch changes in what was heard were associated with greater differences in prosody in individuals with ASD and their parents. Overall, these results highlight how differences (even if very subtle) in the ability to integrate what is heard and said could be a marker of ASD genetic influence, and may be a source of some of the language challenges in ASD.



Do visual attention patterns in ASD run in families?

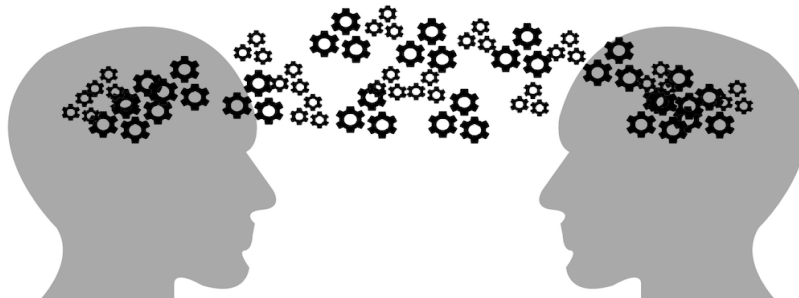
Previous work suggests that individuals with ASD might spend more time looking at objects, such as technology (e.g., computers) and transportation (e.g., trains), and less time looking at people. Time spent looking at people may run in families of individuals with ASD and have a genetic influence. In research presented at the annual scientific meeting for the International Society for Autism Research, we found that a subgroup of parents tended to look more at objects and less at people compared to parent controls. This suggests that looking patterns may run in families of individuals with ASD and be related to language and personality.

Redefining the perception of autism in the workplace

The NDL welcomes a new intern to the lab this summer! An Evanston local, Ira has been extensively involved with the lab for several years since he first participated in one of our studies. Ira always has insightful information to share with us – from playlists of music he enjoys to anecdotes from his experience living with ASD. Ira lives independently and holds a full time position in the grocery business in addition to his internship with the NDL. He is well-established in his professional career, but it hasn't always been easy to get to where he is today. During his early jobs after college, Ira struggled to balance new challenges of adhering to a schedule, interacting with customers, and living away from his family. As he got more accustomed to his routine, Ira was able to successfully navigate these changes and now says his jobs in retail have taught him professional discipline. During his time at the NDL, Ira says he has most enjoyed working alongside the members of our lab and having access to his own office space.



He has found his strengths of organization and attention to detail to be essential to his work in the lab. When asked what he would share with other young individuals with ASD hoping to break into the workforce, Ira emphasized patience and exploration, noting that “not everyone is made to do the same things,” and that taking the time to find a job that is personally interesting will pay off over time. We are so thankful for Ira's contributions to our lab, and for his time with us over the summer!



Unique social-cognitive profiles in ASD and fragile X syndrome

In work led by Lauren Bush, a doc student in the NDL, results were recently presented at the international Society for Autism Research from a study examining theory of mind in children with autism, fragile X syndrome (with and without co-occurring ASD), Down syndrome, and typical development. Theory of mind refers to our ability to understand and predict thoughts and feelings, and that those mental states in others can be different than our own. In this work we found that boys with autism and fragile X syndrome with autism showed a similar pattern of theory of mind abilities, but that children with fragile X without ASD looked more like children with Down syndrome or typical development of a similar mental age. This finding is important because it suggests another important area of overlap in autism and fragile X (in addition to social communication, *see Story time! On p.4*), and also that only a subgroup of individuals with fragile X share this pattern, which is relevant for considering intervention plans. Specifically, we found that boys in both ASD groups (ASD, and fragile X + ASD) showed a unique pattern of passing some developmentally more challenging tasks but having trouble with theory of mind tasks that are usually passed at younger ages. Importantly, we found that this unusual pattern was related to differences in expressive vocabulary (i.e., being able to look at pictures and identify the word the picture is representing), but not receptive vocabulary (our understanding of words) or mental age. This unique pattern detected in this preliminary work might be important for understanding different trajectories and challenges in the development of social understanding that are associated with ASD, which might inform future treatments and interventions.

13th Annual Pepper Lecture

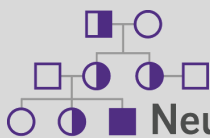
NDL Director, Dr. Molly Losh was invited to give a lecture about our work at the prestigious 13th Annual Pepper Lecture. Roxelyn and Richard Pepper, Northwestern University alumni, endowed the Department of Communication Sciences and Disorders in 2005. The generous gift provided substantial support for clinical and research establishments, training, and research at Northwestern. Dr. Losh's lecture, "Unlocking the Basis of Autism through Studies of Families and Links to the X Chromosome", provided an bird's eye view of our work on autism and related neurodevelopmental conditions. Dr. Losh covered topics that addressed social communication and associated abilities and how such features may span diagnostic boundaries in ways that are critical to understand for diagnosis and treatment, and help to bridge gaps between observable clinical behaviors and underlying biology—a critical step towards understanding the causes of autism and related conditions. Her lecture also focused on research findings from the NDL that can help us understand the boundaries and overlap between disability and unique skills that are tied to genes we all carry, and how genes involved in autism, fragile X, and related conditions can help us to understand genetic influences on complex traits in all of us.



Story time!



Individuals with ASD and fragile X syndrome often have difficulty with social communication skills, which can be evident in their storytelling and other language abilities. In her dissertation research in the NDL, Michelle Lee examined storytelling, conversation, and other language-related skills to understand the picture of the social communication skills in boys with ASD, fragile X, Down syndrome, and typical development. Results showed that there were key similarities between the profiles of the boys who had ASD and those who had both fragile X and ASD, especially in conversational interactions where the ability to interact with another person is strongly tapped. This overlap between the groups suggests that the *FMR1* gene involved in fragile X plays a role in the development of social communication challenges related to ASD. These findings also point to the importance of assessing ASD in children with fragile X, so that most effective treatment options can be tailored for the unique social communication challenges of this group.



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Research supported by grants from the National
Institutes of Health (R03MH079998-01, NIMH;
1R01DC010191-01, NIDCD; 1R01MH091131-01A1,
NIMH), National Science Foundation, and Autism Speaks



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