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! It's been awhile since our last update but we have been hard at work and have many exciting new results to share with you. Most importantly, though, we want to thank you for your participation in our studies. Family support and participation are the most important ingredients to our research, and your support has resulted in a number of important findings emerging from our ongoing projects. We also have several new NIH-funded projects that we hope you will consider learning more about 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!

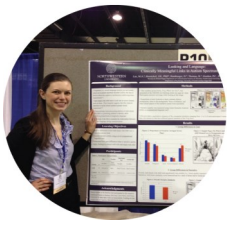
Overview of Our Research

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.

Fragile X and Autism

Fragile X syndrome is caused by a mutation of the *FMR1* gene on the X chromosome, and is the most common inherited cause of intellectual disability. Many individuals with fragile X are diagnosed with autism. Understanding the overlap in language and behavioral features of individuals with fragile X and their families and individuals with autism and their families presents an important opportunity to explore the relationship between genes and behavior, and to discover how *FMR1* might interact with other genetic and environmental factors to cause symptoms of autism. As our research progresses, our goal is to produce findings that can be translated to better treatment and assessment for fragile X and autism.

Highlights



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!

Neurodevelopmental Disabilities Lab
ndl.northwestern.edu | 1-877-275-7187

Check out our website for copies of recent papers!

UNC to St. John's

Dr. Gary E. Martin

Dr. Gary E. Martin, a speech-language pathologist, researcher, and long-time collaborator of the NDL, recently joined the faculty of St. John's University in the Department of Communication Sciences and Disorders. In his new position, Dr. Martin continues to pursue research focused on the speech and language characteristics of children with fragile X syndrome, autism, and Down syndrome. He also teaches courses in the Department of Communication Sciences and Disorders.

st. john's UNIVERSITY

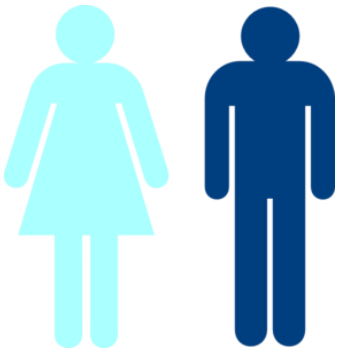
New Findings

Parents' Childhood Development: A Window into Autism Risk?

In a 5-year study funded by the National Institutes of Health, we examined a very valuable and unique resource – archived standardized test scores from parents of individuals with autism, from grade-school through high school. We studied patterns of academic growth in parents when they were children, to learn whether profiles of academic performance over childhood might mark genetic risk to autism. Our findings showed that overall, parents performed significantly above norms in the domains of language, reading, and math. When we looked at patterns of variability across these skills in parents, we found that patterns of development were related to certain personality features in adulthood (e.g., social reticence), and also to the severity of autism symptoms expressed in the next generation. In particular, we found that parent-child associations were specific to parents' childhood language abilities, but not their reading or math abilities. For parents of children with autism, differences in language testing were associated with greater autism severity in their children. We also found that the severity of repetitive behaviors in children with autism was associated with bigger gaps in parents' childhood language, reading, and math skills across development over time (where certain skills developed more rapidly than others). These important findings will help us better understand how certain abilities may relate to genes in autism, and may help us to understand and predict autism genetic risk in families.

Gender Differences in Autism and Fragile X Syndrome

Most of the research conducted about autism and fragile X syndrome focuses on males since there are more males than females diagnosed with both disorders. This focus on males means that we could be missing important information unique to females with autism or fragile X syndrome. We are currently investigating strengths and weaknesses in language, perspective taking, and social skills that may be different between boys and girls with autism, fragile X syndrome, Down syndrome, and typical development. Our early findings show that girls and boys with fragile X do indeed have different social language profiles that are important to consider in educational and intervention planning -- stay tuned for more!



Understanding the Clinical Overlap of Autism and Fragile X: Implications for Intervention and Services

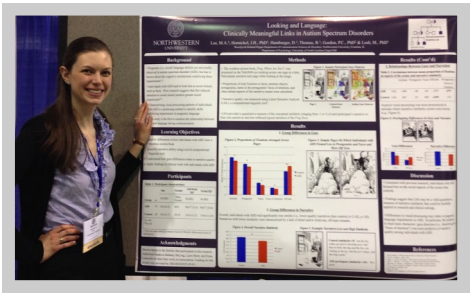
Though many children with fragile X syndrome are diagnosed by a clinician as also having autism, little is known about the consistency between clinical diagnoses and research-based diagnostic measures. This is important because many children with fragile X do not receive clinical evaluations for autism, and therefore miss out on important therapies that could be effective for particular symptoms unique to children who have both fragile X and autism. This study examined whether boys and girls with fragile X met diagnostic criteria on gold standard diagnostic measures in a research setting, and compared rates of research diagnoses to rates of parent-reported clinical diagnoses. Approximately 75% of boys and 25% of girls in the study met full criteria for autism. In contrast, only roughly 25% of both boys and girls had previously received a clinical diagnosis of autism. This study confirms that autism is under-diagnosed in children with fragile X in clinical settings, suggesting that many children with fragile X may be missing out on opportunities for more effective therapies.

Subtle Traits Reflecting Autism Genetic Risk Differ in Fathers and Mothers

Previous studies have identified subtle language and personality traits among family members of individuals with autism that may be linked to the genes that cause autism. In a recent study we looked at family units and how these language and personality traits might differ in mothers and fathers. While traits were evident in both moms and dads, social-personality features like social reticence, or aloof personality, were more common in dads. Paternal personality traits were also related to the severity of children's autism symptoms, particularly in the area of communication. These findings suggest that social personality traits, and differences between mothers and fathers of children with autism, may be markers for how the underlying genetic risk factors for autism are inherited. Findings also have important implications for how the genes that cause autism might also play a role in personality traits (often associated with high achievement and unique skills) in all of us.

Research Presentations

Eye Tracking Reveals Differences in Visual Attention Associated with Language Impairments in Autism



Many studies have shown a link between "looking and speaking". Where our eyes move and stop, and how much time we spend focused on different features of our environment can reveal much about how we are processing the world around us. Individuals with autism have been shown to attend less to emotionally relevant features of scenes, and also show differences in micro-level eye movements in social and nonsocial environments. In a series of studies we have investigated how such differences may relate to language differences in autism, and whether eye movement in parents may also relate to complex language and personality traits that might be linked to the genes involved in autism.

In a study led by doctoral student Michelle Lee, we examined the link between visual attention and complex language measured through narrative storytelling tasks. We found that individuals with autism process both social and nonsocial scenes differently than those without autism, and that these differences relate to the quality of their language. Together, these results provide evidence that differences in visual attention may contribute to social communication difficulties in autism. Findings may have important implications for new interventions in autism to support social communication skills.

Awards:

Lauren Bush

Lauren Bush received a student Travel Award to attend the International Meeting for Autism Research in May 2016. Lauren's research identified key areas of pragmatic language that might serve as valuable markers of genetic risk to autism.

Eye Tracking Reveals Further Insights into Language Processing Differences in Autism

In another study using eye tracking, presented at the International Meeting for Autism Research by doctoral student Kritika Nayar, we examined eye movement during a language processing task, which involved naming different colors, symbols, and objects. The connection between eye movement and naming speed and fluency is a reflection of underlying language processing style, and taps a wide network of brain regions that also support complex social language use. This task was administered to both individuals with autism and their parents, as well as control participants without autism or a family history of autism. Results indicated individuals with autism demonstrated less automated connection between eye movement and language compared to controls, and that this was related to slower language processing, as well as impairments in social language use. We also found that in parents, very subtle differences in eye movement were associated with variation in language processing ability. These patterns were qualitatively similar but much more subtle with no evidence of impairment. Overall, results demonstrate that reduced automaticity in language processing, evident through studies of looking and speaking, is associated with downstream language skills, emphasizing automaticity as a critical precursor to complex language skills and a potential indicator of genetic liability to ASD.

SRCLD 2016

SYMPOSIUM ON RESEARCH IN CHILD LANGUAGE DISORDERS

UNIVERSITY OF WISCONSIN-MADISON

Social Language Development in Boys and Girls with Down Syndrome

Structural language deficits are a prominent feature of the language profile in Down syndrome, leading to a complex pragmatic, or social language, profile where social motivation is strong but language competence may lag. We recently completed a study of pragmatic language development in children with Down syndrome, using a longitudinal, multimethod approach which included three different methods: a parent questionnaire, a standardized measure of social communication, and clinical-behavioral ratings of communication. In findings presented at the Symposium on Research in Child Language Disorders by Michelle Lee and in a manuscript under review for publication, we found that children with Down syndrome demonstrated greater difficulty than younger typically developing controls on parent report and standardized assessments, but only girls with Down syndrome differed on direct assessments. These differences were not accounted for by differences in structural language or cognition. Results suggest that while girls with Down syndrome may show similar pragmatic challenges as boys with DS, when compared to their typically developing female peers, they show greater difficulties with social communication. This finding is very important for considering the intervention and educational supports that may differ between boys and girls with Down syndrome. We also found that the ability to understand others' thoughts and emotions, and executive functioning skills like behavior inhibition and memory related to pragmatic abilities in children with Down syndrome, which helps to highlight underlying skills that can be targeted in interventions.

In July, the NDL will attend the 15th International Fragile X Conference in San Antonio. We are excited to have the opportunity to meet family members and other members of the fragile X community, and also re-connect with families who have previously participated in our research. Our lab will be presenting new findings, and we will also have many opportunities for families to participate in our studies. Our hope is that this may be more convenient for families than traveling to Chicago or arranging our travel to you! Please contact us if you might be interested in participating in our research when we are in San Antonio and we can share more information about the studies, compensation, and make sure you don't miss any of the conference!

New Findings to be Presented at the Fragile X Conference

Jamie Barstein will present findings from a study examining the role of autism symptomatology on communicative repair skills in boys and girls with fragile X syndrome. That is, if a listener indicates misunderstanding (e.g., "what?", "what do you mean?"), what strategies does a child with fragile X syndrome use to attempt to clarify or repair their original message? This is an important skill to study since children with speech and language difficulties may often be asked to clarify messages, so a better understanding of these skills may highlight important new areas for intervention to facilitate communication skills. Strategies were compared to children and adolescents with other developmental disabilities (autism, Down syndrome) and typical development. Results suggest that boys with fragile X syndrome have difficulty using effective repair strategies, with most profound differences noted in boys who also have autism. These findings suggest that autism symptoms significantly affect the communication skills of boys with fragile X syndrome and highlight the importance of teaching effective strategies to repair conversational breakdown.

Michelle Lee and Lauren Bush will present findings from their study examining relationships between executive functioning (e.g., memory, attention), anxiety, depression, and social-personality features in women who carry the *FMR1* premutation. Preliminary results suggest that a subgroup of women report experiencing significant anxiety, depression, and executive functioning difficulties. We also found that these symptoms were related to social cognition, social communication, and personality styles, highlighting the complex interrelationships between psychiatric symptoms, and social and communicative behaviors that may be influenced by the *FMR1* premutation. These results may lead to improved understanding of the psychological, cognitive, and language features associated with the *FMR1* premutation, and have an impact on the design and implementation of targeted supports.

Dr. Gary E. Martin will present findings from a study examining conversational skills of boys and girls with fragile X syndrome with and without autism, as well as children with autism without fragile X syndrome, Down syndrome, and typical development. Boys and girls with fragile X syndrome and autism, and boys with autism-only used more noncontingent (off-topic or tangential) language and perseveration (excessive self-repetition) than other groups, which points to a specific role of autism in the social language impairments observed in individuals with fragile X syndrome. On the other hand, boys with autism-only also initiated less often and were more non-responsive than other groups, whereas boys with fragile X syndrome and autism did not show similar deficits in initiations and responses, suggesting a qualitative difference in social language profiles between the two autism groups. However, girls with fragile X syndrome and autism were more non-responsive than boys. Together, findings indicate that both autism status and sex should be considered in assessment and intervention efforts.

NDL in the Community

Helping High School Students with Disabilities Obtain Jobs

Project SEARCH Intern, Marice (left) meets with Dr. Molly Losh

Our lab recently partnered with Project SEARCH, a program that is designed to help high-school students with autism prepare to transition into the workforce through internship experiences at Northwestern University. Since the project's launch in fall 2013, the NDL has had the privilege to work with many different interns. Each served as valuable members in our lab, taking on a variety of responsibilities ranging from daily clerical duties to detailed data entry on the computer. With oversight from job coach TC Schneck, the interns were able to gain important job skills and excel in their duties. While each internship placement lasts one quarter, it is evident that the program provides many benefits. Program Coordinator Sara LaMontagne explains, "While at the Neurodevelopmental Disabilities Lab, each intern gained many crucial employability skills that will help them transition into the working world. They learned a variety of data entry skills during their time as an intern in the lab. Thanks to the Neurodevelopmental Disabilities Lab, they have made incredible improvement on their job and social skills and are one step closer to gaining meaningful employment after Project SEARCH!"

We have loved working with the interns in the lab, and appreciate all of their hard work – each individual provided invaluable assistance to our research projects!

For more information about Project SEARCH, visit www.projectsearch.us; or contact familystudy@northwestern.edu

Let 'Em Know!

July is Fragile X Awareness Month. In 2015 the NDL teamed up with the Heartland Fragile X Alliance to fundraise and help spread awareness for Fragile X by participating in the National Fragile X Foundation's Let 'Em Know 5k Run/Walk event.

Greater Chicago Fragile X Research Event at Rush University Medical Center

Dr. Molly Losh pictured with Dr. Berry-Kravis, and members of the NDL, Erica Karp and Nell Maltman

In April, Dr. Molly Losh was a featured speaker at Rush University Medical Center, along with Dr. Joanne O'Keefe (Rush), Dr. Deborah Hall (Rush), Dr. Anne Hoffman (Rush), and Dr. Elizabeth Berry-Kravis. The purpose of this event was to update families on new findings from our studies, and share new study opportunities for families. This terrific event was organized by Missy Zolecki of the Greater Chicago Fragile X Group. We were grateful to be a part of it and so enjoyed connecting with families who attended!

Autism Speaks Midwest Employment Symposium

In March 2016, in partnership with Northwestern's Center for Audiology, Speech, Language, and Learning, our lab co-sponsored the Midwest Employment Symposium at Northwestern University. The day-long event brought together people with autism, families, researchers, business owners, entrepreneurs, and stakeholders and community members to help address employment issues for adults with autism. The action-packed event included panel discussions, employee presentations, business competition, a resource fair, and audience Q&A.

The event featured several businesses that employ people with autism, including:

- 100% Wine, a St. Louis-based company that donates all profits to nonprofit organizations working to create jobs for people with a disability.
- Highland Park's Aspirtech, which provides domestic software testing services.
- Sugar and Spice Extraordinary Sweet Treats in Evanston, which hires young adults with disabilities and runs a workplace training program for people with autism.

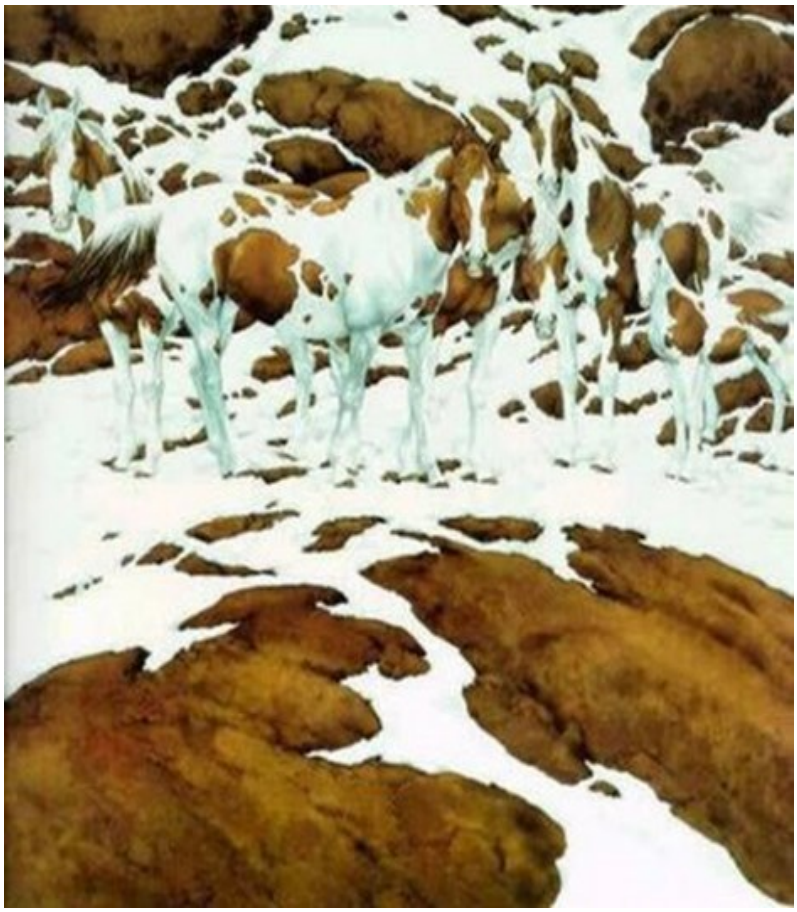
Awards:

Kritika Nayar

Kritika Nayar, a second year doctoral student in the lab, has been awarded the Northwestern University Cognitive Science Advanced Fellowship. Kritika's work examines visual perception in individuals with autism and their parents in relationship to underlying electrophysiology. The work aims to foster cross-departmental collaboration with Dr. Satoru Suzuki from the Department of Psychology and will take a transdisciplinary approach to assessing underlying cognitive styles of visual perception associated with autism using neural and behavioral measures.

Looking Ahead: New Studies

Visual Perception Styles: A Family-Genetic Study



Assessing Perceptual Styles: How many horses do you see in this image?

We recently received funding from the National Institutes of Health for a new study assessing perceptual skills in individuals with autism and their relatives. Visual processing enables us to perceive features of our surroundings and integrate them into a coherent whole. Differences in perceptual styles can significantly influence how we interpret our physical and social worlds. We are interested in understanding how individuals with autism attend differently to scenes (e.g., “seeing the trees before the forest" or "seeing the forest before the trees"), but also how these abilities may have a genetic basis. Using eye-tracking methods to examine visual perception during multiple computer-based tasks, the goal of this study is to provide knowledge about underlying processing strategies which may importantly relate to clinical features of autism, and serve as markers of underlying genetic influence related to autism. Please keep a look out for updates on this study!

Studies of Brain Function Related to Language

We are conducting new research to explore the brain-basis of language differences in autism and fragile X syndrome, and whether neural signatures related to language may be heritable within families. These studies use noninvasive measures that allow us to measure neural representations of speech in the brain. During this task, the participant wears three sticker-like electrodes on his or her head and hears some different sounds through earphones. The brain does the rest of the work, so the participant is able to relax and watch a movie during the task. Research has indicated strong links between these neural measures and language, and in this work we aim to identify specific neural markers that can be used to learn more about the biological basis of the language features in autism and fragile X.

Virtual Job Training

NDL members are collaborating on an innovative computer-based intervention (led by Dr. Matthew Smith) to facilitate job skills in adolescents and adults with autism. Using Virtual Reality, this study involves presenting computer-based simulations assessing job interviewing skills, and job-related interactions that vary in level of difficulty, complexity, and intensity, and are designed to simulate the experience of a "real-world" workplace environment. We are excited about this new line of research, and its potential impact on the community.

Intervention Research

The Neurodevelopmental Disabilities Lab recently began collaborating with the Early Intervention Research Group at Northwestern (led by Dr. Megan Roberts) to learn more about the efficacy of parent-mediated intervention. The goal of the project is to discover whether parent personality-related traits are a feature that may be important to consider in planning effective interventions.

The study will better inform future clinicians by maximizing the possible intervention strategies that are best aligned with particular personality and language traits.

Awards:

Jamie Barstein

Jamie Barstein, a PhD student in the lab, received an award from Northwestern University's Alumnae Association. Jamie's work examines the potential link between sensory processing differences (e.g., under- or over-sensitivity, sensory-integration problems) and communication profiles among children and adults who carry the *FMR1* premutation. Jamie is also interested in studying the link between sensorimotor and communication profiles in parents of individuals with autism.

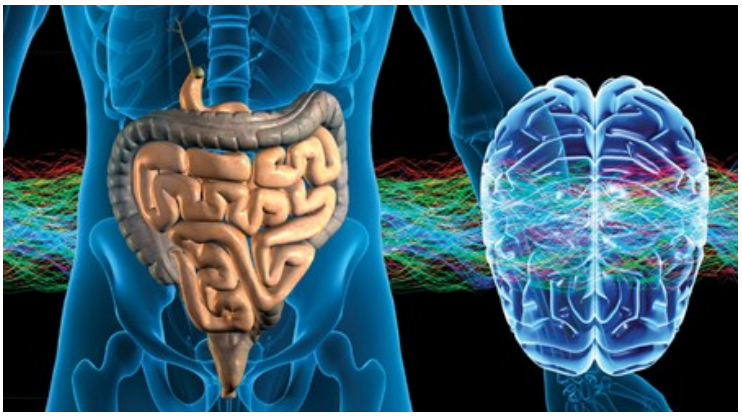
If you went to grade school or high school in Iowa or North Carolina, you probably remember taking standardized tests like the Iowa Test of Basic Skills (ITBS). Iowa and North Carolina are very unique states because they archive individual test scores and will release them for use in research, with the individual's consent. This is an invaluable resource because it provides information about individual abilities throughout development.

Please be in touch to learn more!

Studies of the Connection between Brain and Gut

Research findings show that individuals with autism may suffer from a range of sensory and gastrointestinal issues, and that these symptoms may importantly relate to cognitive function and behavior. Our lab is conducting exciting new research investigating the basis of these issues, and how they may relate to other features of autism, as well as whether these symptoms are heritable and evident in relatives.

Please contact us for more information!



New Technology for Research

We have recently begun studying brand new experimental measures of cognition, language, and social behavior, all employing a participant-friendly iPad, and with shorter tests to ease the burden on participants. These new measures, called the "NIH Toolbox," were developed by researchers funded by National Institute of Health and incorporate multiple areas of neurological functioning, including cognitive, motor, emotional, and sensory functioning. Integrating the NIH Toolbox allows us to quickly and accurately gather this information, giving us more time to focus on our primary research goals, and we are excited to have begun implementing these powerful new tools in some of our research studies.

Dr. Losh and graduate student Jamie Barstein were invited to speak on the implementation of the NIH Toolbox at the Health Measures Users Workshop on June 2nd. Their presentation focused on our lab's experience utilizing a new iPad version of NIH Toolboxes as well as preliminary results from a pilot study of individuals with autism and their parents. Your feedback so far suggests that the Toolbox tasks are fun and quick to complete. Thank you to all who have participated and helped us to evaluate this important new tool!

NIH Toolbox
Assessment of Neurological and Behavioral Function

The NDL is on Facebook!
Follow our page for updates on the lab!
WWW.FACEBOOK.COM/NDLFAMILYSTUDY

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NDL Students in the Spotlight

Shivani Patel, a first-year PhD student in the lab, was awarded a National Science Foundation Graduate Research Fellowship to study how young children process the sounds of language and how this may be different in autism and fragile X. Specifically, Shivani's work focuses on examining prosody, a key component of pragmatic language, in individuals with autism, fragile X syndrome, and their parents. Shivani is currently examining expressive prosody in different contexts, including narrative and conversation. Examining parallel language patterns in affected individuals and their parents is critical for expanding our knowledge of familial patterns of language profiles.

PhD Student Featured in *Breakthroughs*

Jamie Barstein, a second-year PhD student in the lab, was featured in the Feinberg School of Medicine's *Breakthroughs* newsletter. The article, titled *Linking Behavior and Genetics*, highlights Jamie's work in the lab and her journey in pursuit of a doctorate in clinical psychology.

For full article visit: http://www.feinberg.northwestern.edu/Research/docs/newsletters/March2015.pdf

Natalie Dulin, a senior undergraduate research assistant in our lab, is completing an honors thesis on sex differences in pragmatic language in individuals with autism and their parents. Findings thus far do not indicate significant sex differences; however, qualitative trends show that females in both groups have stronger pragmatic ability than males. Natalie was accepted to present her findings at the Northwestern Undergraduate Research and Arts Exposition in June.

Nancy Dunbar recently began a position as a staff member in the NDL. She first started working in the lab as an undergraduate, as a member of the Summer Undergraduate Research Assistants Program in the summer of 2015. She loved her experience so much that she decided to continue working in the lab through independent study during her senior year. After graduating from Northwestern in June, we are thrilled that she has joined our team as a full-time staff member. She is looking forward to being even more involved in of all of the exciting research happening in the NDL!

Neurodevelopmental Disabilities Lab

Northwestern University

Director: Molly Losh, PhD

Key Collaborators:

Elizabeth Berry-Kravis, MD, PhD, Rush University Medical Center

Gary E. Martin, PhD, St. John's University, New York

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NORTHWESTERN UNIVERSITY

NEURODEVELOPMENTAL DISABILITIES LABORATORY

THE ROXELYN AND RICHARD PEPPER DEPARTMENT OF COMMUNICATION SCIENCES AND DISORDERS

2240 CAMPUS DRIVE

EVANSTON, IL 60208