RABLAB NEWSLETTER

UNIVERSITY OF WASHINGTON **WINTER 2017**

Happy New Year from the Bernier Lab!

Hello again and welcome to the Bernier Lab at the UW! First we would like to extend a big thank you to our study participants and wish everyone a Happy New Year! During 2016 the RabLab was very productive: we started several new research studies, published findings from past and ongoing research, and built new collaborations and community partnerships to advance the field of Autism Spectrum Disorder (ASD). This edition of the RabLab Newsletter summarizes several of our recent research findings published in scientific journals, reports RabLab news and events from Fall 2016, and provides information regarding current research studies. Enjoy!

rablab@uw.edu * www.bernierlab.uw.edu
206.616.2889 * www.facebook.com/bernierlab

RESEARCH FINDINGS

Disruption of POGZ is associated with Intellectual Disability and Autism Spectrum Disorders

Disruptive mutations in POGZ have been associated with intellectual disability (ID) and autism spectrum disorders (ASD) in previous investigations. The current study provided detailed clinical and functional information on 25 individuals with disruptive mutations in POGZ. Common clinical features included varying levels of developmental delay (more severe speech and language delay compared to motor delays) with most individuals having mild ID. Strikingly, in several individuals, the ability to speak in sentences and write and read simple language started very late, but was eventually acquired. A distinct neurobehavioral phenotype could be recognized, including either a formal ASD diagnosis or features of ASD and, in many cases, a seemingly contrary overly social and overly friendly demeanor. Associated medical conditions included vision problems, microcephaly, hyperactivity, a tendency towards obesity, feeding difficulties, and sleeping problems. In addition, researchers modeled the POGZ partial loss of function in fruit flies and found evidence for the importance of this protein in cognitive functioning and learning. Although the phenotypes were variable, this study provided evidence of shared phenotypic features, suggesting that mutations in POGZ might represent a distinct ASD and/or ID syndrome. **Stessman, H, et. al, 2016. Am J of Hum Gen, 98 (3):541-552.**

Modulation of mu attenuation to social stimuli in children and adults with 16p11.2 deletions or duplications

These findings compared typically developing individuals and individuals with ASD using mu attenuation. Mu attenuation is computed through Electroencephalogram (EEG) signals recorded while individuals were observing videos of social, nonsocial, or no motion as an expression of social brain phenotype. Mu rhythm is believed to reflect activity of an action execution-observation matching system which plays an important role in many aspects of social cognition. Results indicates that typical individuals show greater mu attenuation for social motion relative to nonsocial whereas both 16p11.2 CNV carriers exhibit greater mu attenuation for nonsocial compared to social motion. Individuals with ASD, however, exhibits equivalent levels of mu attenuation regardless of conditions. By using single-trial analysis, the results confirm that mu attenuation decreases more rapidly for 16p11.2 deletion carriers than either the duplication carriers of the typical group but we see an increase of mu attenuation over time for ASD.

Hudac, C., et. al., 2015. J of Neurodevl Dis 7(1), 25-36.

rablab@uw.edu * www.bernierlab.uw.edu
206.616.2889 * www.facebook.com/bernierlab

Developmental Trajectories for Young Children with 16p11.2 Copy Number Variation

This study explored differences in development between children with a duplication or deletion of a section of their 16th chromosome, called 16p11.2. This genetic region has been associated with ASD symptoms, as well as variable cognitive and behavioral difficulties. 56 children ages 6 months to 5 years were studied across time in terms of their cognitive, adaptive, and behavioral skills. Children with a deletion at 16p11.2 were compared to those with a duplication at 16p11.2. Children with 16p11.2 deletions show a pattern of early gains in verbal cognitive ability alongside losses in motor and social skills during that same time period. In contrast, children with 16p11.2 duplications tended to show early gains in verbal cognitive abilities without losses in motor and social skills. In other words, decline in motor and social abilities early on in development may be characteristic of 16p11.2 deletions and unique compared to other disorders. Duplications to the same genetic region appear to result in steady growth in motor and social domains. Interestingly, children who went on to be given specific psychiatric diagnoses showed differences in their early symptoms presentation compared to children who were not given the same diagnoses. Children with 16p11.2 duplications who were later diagnosed with ASD trended toward greater impairment in cognitive, communication, and social skills but fewer disruptive behaviors compared to those without ASD. These results are promising as they suggest there may be different developmental patterns that occur for 16p11.2 deletions and duplications, which could help inform more targeted treatments early on in child development to potentially improve outcomes over time.

Bernier, R. et. al, J. or Med Gen, Part B. In Press.

RABLAB NEWS

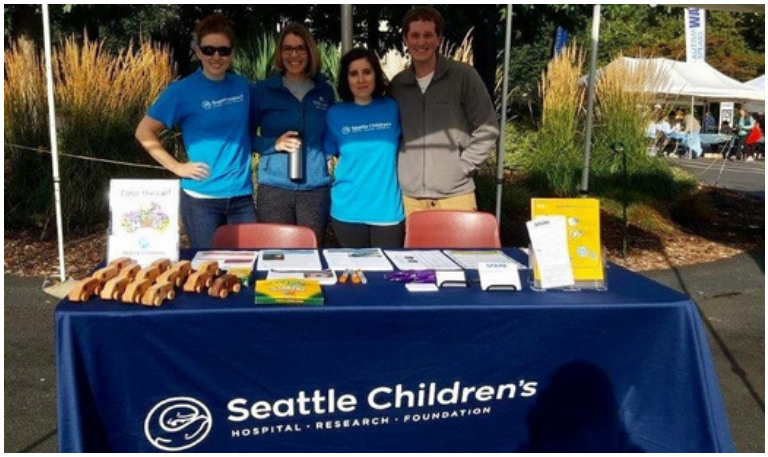
THE EEG SESSIONS

The Rablab's Blog hosts a new series of posts called 'the EEG Sessions,' where members of the Lab's EEG team discuss EEG related topics and articles. Part of these Sessions are the 'EEG journal Club', which consists of the team reading and discussing an article of interest and sharing their thoughts on our blog. One of the goals of the EEG journal club is to think critically about existing research in order to improve our won methods and produce reliable findings with clinical applications.

https://depts.washington.edu/rablab/category/eeg-sessions/

If your address or contact information has changed, please let us know by emailing rablab@uw.edu!

WALK NOW FOR AUTISM SPEAKS



The Puget Sound Walk Now for Autism Speaks was on September 24th! It was held at the Seattle Center's Next 50 Plaza and had over 1,000 attendees. They raised more than $170,000 and hope to reach their goal of $196,500 by the end of the year with continuous donations.

Above: Some of our lab staff attended the event and handed out information on the Spark, GABA, and TIGER studies! They also gave out small wooden cars that could be painted, played with, and then taken home by children attending the walk! To make a donation to Autism Speaks, go to www.autismspeakswalk.org/pugetsound!

LAB MEMBER SPOTLIGHT

Jessica Peterson, PhD

Jessica Peterson, Ph.D., is a post-doctoral fellow with the Bernier Lab. Dr. Jessica is passionate about providing services to individuals with autism spectrum disorder and their families as well as researching factors that advance our understanding of autism and inform interventions to improve outcomes and quality of life for families.

What do you like most about working in the Bernier Lab?

The most amazing part of my job is working with families who participate in our research and the Bernier Lab staff. I love meeting and working with new families every week and learning about their unique experiences over a child's lifetime, how they found their way to our lab, and what we can do to support families of children with autism and related disorders in the future. The Bernier Lab staff are some of the most caring, enthusiastic, and effective researchers around which makes it exciting to be at work every day.

What makes the "genetics-first approach" to ASD research so important for you?

The "genetics-first" approach is really important to me especially when I think about the level of differences, the huge variability we see in individuals with autism and the need to better understand these differences. I am grateful to be part of a team that is working to understand and connect genetics with unique autism profiles and use this information to personalize treatments and improve quality of life for families. I really enjoy the opportunity to provide families with the new science and information that we know thus far from our lab.

Follow and like our official Bernier Lab Facebook page at **facebook.com/bernierlab**, and check out our website at **www.bernierlab.uw.edu** for up-to-date study information and family resources!

CURRENT RESEARCH

ABC-CT

The Autism Biomarkers Consortium for Clinical Trials (ABC-CT) is seeking families to participate in a study to improve diagnosis and treatment in Autism Spectrum Disorder (ASD). We are currently recruiting both typically developing children and children with a diagnosis of ASD between the ages of 6 and 11 years old. The aim of the consortium is to develop reliable and objective measurements of social function and communication in people with autism.

GABA

We are currently looking for adults ages 18-30 both with and without ASD to participate in a study of how the brain processes sensory information. Several MRI scans are required. Additionally, we have expanded to recruit children ages 10-14!

SFARI SPARK

The goal of the SPARK project is to collect genetic information on 50,000 individuals with ASD and their families across the country. Anyone with a diagnosis of autism can participate. To register for SPARK, go to www.SPARKforautism.org/UW or contact 206-987-7917 | SCACstudies@seattlechildrens.org

TIGER

We have seen over 80 families so far! We are still looking for children ages 4 and up for a comprehensive study of particular genetic events associated with ASD.

TWINS-2

We are looking for identical twin pairs ages 4 and up in which at least one twin has ASD to participate in a genetic study. In person visits to our lab are not required for participation, so families worldwide are welcome!

ZEBRA

We have seen over 125 families so far! We are still looking for children ages 8-17 in the greater Puget Sound area for an EEG study of brain mechanisms and behavior in ASD.

If you are interested in participating in any of our studies, please contact us at (206) 616-2889 or rablab@uw.edu