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the

RABLAB NEWSLETTER

UNIVERSITY OF WASHINGTON

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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! We have been busy at work at the Lab seeing families for our current studies, publishing new research findings, sharing science through video blogcasts, and collaborating with our local community to build public awareness and capacity to support individuals with ASD and their families.

Upcoming Events

March 15, 2018

Seattle Children's

Autism 203: Social Inclusion through Person Centered Planning

April 19, 2018

Seattle Children's

Autism 204: Powerful Partnerships: Strategies for Navigating the Family/School Relationship

May 17, 2018

Seattle Children's

Autism 205: Inclusion: What's Working and What's Next? – A Panel Discussion

RABLAB NEWS

It's Showtime: Autism Anchors

Join our very own Dr. Bernier, along with Jim Mancini, SLP, each month on Spectrum News (https://spectrumnews.org/features/special-report).

From science to the community and back again, you can count on getting the inside scoop in digestible quantities with a smile from the autism anchors.

Attention and ASD

Congratulations to our post-doctoral fellow Dr. Anne Arnett, whose paper on inattention and ASD comorbidity was included in the prestigious Autism Science Foundation's 2017 year-end research review! Check out a brief summary of her findings below, or read the full research review online at: http://autismsciencefoundation.org/key-autism-research/.

Welcome Inclusion Initiative: WIN

Dr. Raphael Bernier has been a leader in the new Welcome Inclusion Initiative, which hosted its first symposium on June 27, 2017.

The Welcome Inclusion (WIN) initiative is a grassroots alliance. WIN is a public awareness and capacity building campaign that will facilitate rapid, transformative community change to promote a world of inclusion for children and adults with intellectual, behavioral, and social differences.

https://www.welcomein.org/

Professorship

Congratulations to Dr. Raphael Bernier, who was promoted to full Professor in the Department of Psychiatry and Behavioral Sciences!

SPARK PIRATE DAY!
@ SEATTLE CHILDREN'S AUTISM CENTER

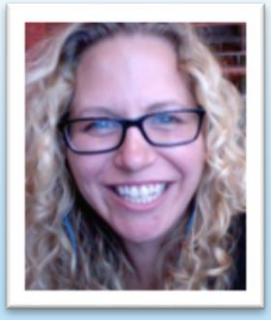
Our research team hosted a pirate-themed, in-person research day on Saturday, December 2nd at the Seattle Children's Autism Center! We provided families with snacks, games, and childcare while they participated in the SPARK study and in our new genetics research study, PANGEA. Kids wore pirate hats, colored a gigantic pirate ship, searched for buried treasure, and picked prizes out of a treasure chest.

With an all-hands-on-the-[pirate]-deck approach, we completed **18 families for SPARK**. This was also the first day for a **new blood-draw room** at the Seattle Children's Autism Center. For families interested in participating in PANGEA, they will now be able to complete their blood draws at the Autism Center! On Research Day, we completed **43 individual blood draws** for the PANGEA study! We are hoping to do another fun event in the spring, so stay tuned for details!

Lab Member Spotlight: Caitlin Hudac, Ph.D.

About Caitlin

I am a 4th year postdoctoral fellow. I've been working in autism research for 10 years! Starting in 2007, I worked with Dr. Kevin Pelphrey using fMRI to learn more about the social brain. I did my doctoral training at the University of Nebraska, Lincoln (Ph.D., '14) where I used EEG and ERP to learn about how newborns and infants begin to think about people. I've been in the Bernier lab since 2014 and most of my efforts involve looking at biomarkers of attention and social cognition. Specifically, I'm proud of our efforts in specifying patterns of habituation in children with genetic events, such that we see unique patterns regarding how information is processed and how brain signals reduce over time.



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

Each person in the lab views autism from a different lens — we look at medical history and symptoms, clinical presentation, behavior, body shapes and sizes, and some of us (like me) look at the brain. Together, we see the whole picture and I am confident that we are making progress in developing biomarkers that will help with clinical interventions. Plus, the PI does some really killer Duran Duran karaoke.

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

Autism is so complex and diverse — using a genetics-first approach allows us to dig into (potentially) unique and distinct types of autism. It is incredibly inspiring to connect with families and hear stories about daily life. Each conversation and each time we work with a different child, I see a new feature or another puzzle piece which helps us bridge the similarities, so we can better understand how the brain works.

Q: What is your favorite thing to do in Seattle?

More often than not, you'll find me on my bike, heading to a coffee shop, bonfire, karaoke, or catching the ferry to go bike camping.

RESEARCH FINDINGS

Clinical Phenotype of ASD-associated DYRK1A Haploinsufficiency

Recurrent disruptions to the *DYRK1A* gene are seen in as many as .1 to .5% of individuals with ASD. Historically, ASD research has focused on categorical descriptions of phenotype prior to genome sequencing. Bernier Lab has employed a new approach, exploring phenotype with respect to the *DYRK1A* gene. Results confirm a distinct phenotype for *DYRK1A* disruptions which includes speech delay, anxious behaviors, feeding difficulties in infancy, and microcephaly (decreased head circumference.) Extensive effects on facial structure, such as deep-set eyes and a high nasal bridge, have also been noted. Results suggest a potential subtype of a recurrent disruptions to the *DYRK1A* gene are seen in as many as .1 to .5% of individuals with ASD. Historically, ASD research has focused on categorical descriptions of phenotype prior to genome sequencing. Bernier Lab has employed a new approach, exploring phenotype with respect to the *DYRK1A* gene. Results confirm a distinct phenotype for *DYRK1A* disruptions which includes speech delay, anxious behaviors, feeding difficulties in infancy, and microcephaly (decreased head circumference.) Extensive effects on facial structure, such as deep-set eyes and a high nasal bridge, have also been noted. Results suggest a potential subtype of ASD related to *DYRK1A* disruptions. Nonetheless, severity of impairment caused by *DYRK1A* disruptions varies greatly, likely due to differences in parental phenotype related to *DYRK1A* disruptions. Nonetheless, severity of impairment caused by *DYRK1A* disruptions varies greatly, likely due to differences in parental phenotype.

Earl, RK, Turner, TN, Mefford, HC, Hudac, CM, Gerdts, J, Eichler, EE, Bernier, RA. (2017). Clinical Phenotype of ASD-associated DYRK1A haploinsufficiency. Molecular Autism. 8:54.

Comorbid Symptoms of Inattention, Autism, and Executive Cognition in Youth with Putative Genetic Risk

Autism spectrum disorder (ASD) and the inattention symptom (IA) domain of attention deficit hyperactivity disorder (ADHD) have each been well studied in regards to cognitive deficiency, where executive functions such as inhibition, planning, and flexibility are key endophenotypes of each. While studies have shown that there is a fairly high comorbidity rate (20%-50%), literature on the two disorders have continually had mixed profiles and symptoms often as a result of limitations such as inclusion of only high functioning individuals, the use of dichotomous diagnostic categories which decreases the ability to estimate covariance, and the struggle to find single genes that have a large effect on the development of ADHD. This study sought to find a middle ground of covariance between the symptoms of ASD, IA, and executive dysfunction. A final sample size of 73 individuals ages 3 - 22 years old who have been identified to have a gene disrupting mutation previously associated to ASD were recruited to participate in various tests and batteries including genetic tests, ability scales, verbal and vocabulary tests, among many more while parents filled out questionnaires. The study found that 31% of those that were diagnosed with ASD (89%) also showed Elevated IA symptoms and that IA severity was still higher than the normative population. 11 genes accounted for about a third of the variance between SRS-2 (a measure for ASD symptoms) and IA, which explains the substantial covariance across ASD and IA symptom severity. These genes should be targets of future studies of ADHD while the genes identified to have no correlation to IA should be considered for future ASD studies.

Arnett, AB, Cairney, BE, Wallace, AS, Gerdts J, Turner, TN, Eichler, EE, Bernier, RA. (2017). Comorbid symptoms of inattention, autism, and executive cognition in youth with putative genetic risk. J Child Psychol Psychiatry.

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!

PANGEA

We are currently looking for individuals of all ages with ASD and their families to participate in a study to aid in gene discovery. Participation involves a blood draw and answering brief family history questions. Involvement does not require an in-person visit.

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 are recruiting 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 are 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