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CELEBRATING AUTISM AWARENESS MONTH

The Seaver Autism Center celebrated Autism Awareness Month in April by spreading knowledge about autism and autism research, sharing stories from families affected by autism, and showing our appreciation for the families that participate in important research at the Seaver Autism Center.

#SeaverStories: Each week in April, we shared a #SeaverStory from a family affected by autism, or from a researcher working in the field of autism. This year, the Whitney family discussed their journey to discover their son Jonah had a rare genetic mutation (read the full story on page 5). The Clayton family discussed how they found hope in research for their



The Clayton family shared how they found hope in research for their son Gavin (left) as part of their #SeaverStory.



Dr. Reichenberg discussed how he became involved in autism research as part of his #SeaverStory.

son Gavin. Julia George-Jones talked about her role as a Seaver research coordinator and how it has informed her career plans. Lastly, Dr. Avi Reichenberg discussed the

series of circumstances that led him to his valuable work researching the environmental risk factors behind autism. Read more stories at <http://icahn.mssm.edu/research/seaver/patients/stories>.

Family Appreciation Day: The work that the Seaver Autism Center does would be impossible without the families that participate in research and trials. To show our appreciation, we hosted these families for a day of activities and games with Seaver Center faculty, students and staff.

Continued on page 2

RESEARCH UPDATES

• **Grandma Knows Best:** Children who have frequent interaction with grandparents are diagnosed with autism earlier than those who do not, according to new research conducted at the Seaver Autism Center and published in the journal *Autism* by senior author Joseph D. Buxbaum, PhD. The study's findings suggest that by tapping into the feedback and wisdom of family, friends, and caregivers, it is possible to diagnose autism earlier and therefore improve treatment outcomes.

• **First Rat Model for Phelan-McDermid Syndrome:** Researchers at the Seaver Center generated and characterized a genetically modified rat model for Phelan-McDermid syndrome, a developmental disorder with high rates of autism, intellectual disability, attention deficits, and severe language delay. The study results, published in *eLife* by lead author Hala Harony-Nicolas, PhD, Instructor at the Seaver Autism Center, provide important leads into how the specific mutation

mimicked in the rats plays a role in synapse development and function and, ultimately, behavior. The study also found that the hormone oxytocin significantly improved social memory, attention, and nerve cell activity, which may help inform novel treatments.

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AUTISM SEQUENCING CONSORTIUM AWARDED \$7 MILLION IN RENEWED FUNDING

The National Institute of Mental Health recently renewed its support of the Autism Sequencing Consortium, an international autism genetics research consortium, with an award of \$7 million.

The Autism Sequencing Consortium was founded by Joseph D. Buxbaum, PhD in 2010 with pilot funding from the Beatrice and Samuel A. Seaver Foundation to bring together investigators from around the world to collect and share samples and

genetic data from individuals diagnosed with autism. In 2013, the National Institute of Mental Health awarded a large grant to the ASC to expand the project to include data from more than 20,000 individuals over three years. The Consortium now includes more than 40 international groups and over 150 researchers, who have generated gene sequencing data from 29,000 individuals, making it the largest sequencing study to date in autism, and even in psychiatry. The new grant will extend the project through

2022 and expand the sample to include more than 50,000 individuals.

“Historically, the number of risk genes found has steadily increased with the number of patients studied, so it’s important that we continue to add patients to the data set,” says Dr. Buxbaum. “We are thrilled to receive this grant, which will enable our unique, collaborative research consortium to continue the work that is accelerating such important discovery.”

AUTISM AWARENESS MONTH

Continued



Julia George-Jones, Research Coordinator, and Savannah Lennertz, Marketing & Communications Associate, at an Autism Awareness Month information table.

• **Spreading Awareness:** To continue our mission of spreading awareness about autism and autism research, the Seaver Autism Center hosted a Twitter Chat in partnership with the Autism Science Foundation (find our chat on our Twitter page @SeaverAutism, chat hashtag #AutismScience). In addition, Center Director Joseph D. Buxbaum, PhD answered questions about autism on Facebook Live (find the full video on our Facebook page @SeaverAutismCenter). The Seaver Autism Center also organized information tables throughout the month to answer questions from families and physicians passing through the Mount Sinai lobby.

SEAVER IN THE COMMUNITY

Seaver Research Coordinators and staff played interactive games with local students and families to teach them about autism at two recent community events: Family Science Night at a local public school and the 5th Annual Mount Sinai Brain Fair.



Local students solve a brain puzzle with autism facts at the Mount Sinai Brain Fair.



Seaver Research Coordinators Julia George-Jones, Elyana Feldman and Allison Durkin at Family Science Night.

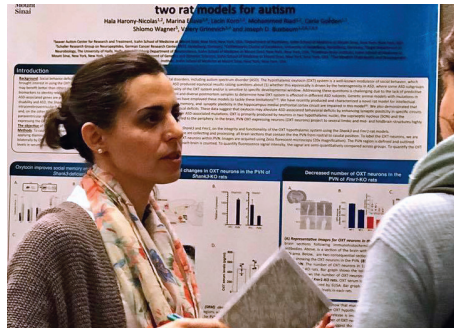
SEAVER RESEARCHERS AT 2017 INTERNATIONAL MEETING FOR AUTISM RESEARCH

At this year's International Meeting for Autism Research (IMFAR) in San Francisco, sponsored by the International Society for Autism Research, a number of researchers from the Seaver Autism Center presented posters on recent research. Poster presenters included Andrew Browne, Elodie Drapeau, PhD, Jennifer Foss-Feig, PhD, Julia George-Jones, Carla Golden, Hala Harony-Nicolas, PhD,

Emily Isenstein, Pilar Trelles, MD, and Ting Wang, PhD. In addition, Avi Reichenberg, PhD, and Sven Sandin, PhD, presented as part of a panel titled "Autism and Intellectual Disability: Patterns of Familial and Environmental Risk." To wrap up the week, the Seaver Center hosted a reception for current and former members of the Seaver Center, as well as friends and collaborators.



Avi Reichenberg, PhD, and Sven Sandin, PhD, (pictured above) presented as part of a panel titled "Autism and Intellectual Disability: Patterns of Familial and Environmental Risk."

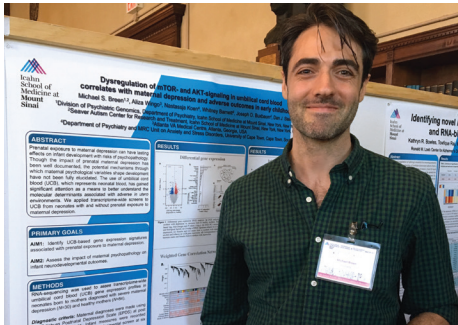


Hala Harony-Nicolas presented a poster titled "Integrity and Functionality of the Hypothalamic Oxytocin System and the Effect of Oxytocin Treatment in Two Rat Models for Autism."



Current and former Seaver Center members, friends and collaborators at a reception hosted by the Seaver Center.

OTHER RECENT PRESENTATIONS



Michael Breen, PhD (pictured above), Andrew Browne, Elodie Drapeau, PhD, Carla Golden, Emily Isenstein, Magdalena Janecka, PhD, and Elisa Nabel presented posters at the 9th Annual Neuroscience Retreat at the Icahn School of Medicine at Mount Sinai.



Allison Durkin and Kristin Meyering, Research Coordinators at the Seaver Autism Center, presented a poster titled "Prospective characterization of four cases of de novo FOXP1 variants" at the Fifteenth Annual Child Health Research Day at Mount Sinai.



Avi Reichenberg, PhD presented a talk titled "Environmental Risk and Genetic Contribution to Autism Risk" at the 10th Annual Winter Symposium hosted by the Children's Environmental Health Center at the Icahn School of Medicine at Mount Sinai.

RESEARCH UPDATES

Continued

• **Paternal Age at Conception May Influence Social Development:** The age of the father at the time his children are born may influence their social development, suggests a study published in the May 2017 issue of the *Journal of the American Academy of Child and Adolescent Psychiatry (JAACAP)* by lead author and Seaver Fellow Magdalena Janecka, PhD. By analyzing social behaviors of children from early childhood until adolescence, researchers found that those whose father was either very young or older at conception differed in how they acquired social skills. These findings may offer insights into how paternal age influences children's risk of autism and schizophrenia.

• **Seaver Postdoc Awarded Autism Science Foundation Fellowship:** Michael Breen, PhD, Postdoctoral Fellow at the Seaver Autism Center, was awarded a Fellowship Grant by the Autism Science Foundation to identify new genes and pathways in blood samples that may improve diagnosis and identify new drug targets that will enhance the development of treatments for autism.

To keep up-to-date on new research from the Seaver Autism Center, follow us on social media or check out icahn.mssm.edu/research/seaver/about/news.

IGNITING COLLABORATION WITH THE CHILDREN'S HOSPITAL OF PHILADELPHIA

In April, a team of researchers from the Center for Autism Research at the Children's Hospital of Philadelphia (CHOP) travelled to New York City to meet with researchers from the Seaver Autism Center at the New York Academy of Medicine. CHOP and the Mount

with the intention of igniting discussion around possible areas of collaboration.

In the afternoon, attendees were challenged to form cross-Center groups to create collaborative proposals that utilized complementary strengths, applied novel approaches, and combined datasets. At the end of the day, each group presented their proposals to a team of "judges," including the Scientific Director of the Center for Autism Research, Robert Schultz, PhD, and the Director of the Seaver Autism Center, Joseph D. Buxbaum, PhD.

"The plans that emerged from this exercise exceeded our expectations," said Dr. Buxbaum. "In just a few hours, our clinical and research faculty identified several real targets for promising lines of research that could begin almost immediately. We each left feeling challenged and inspired by the rich partnership that began to take shape today."

These collaborative teams will continue to refine their projects over the next six months, at which time they will meet again in Philadelphia, with the goal of developing proposals ready to be submitted to the National Institutes of Health as R01 grants.

"Breakthroughs will only come from being able to see the patterns of genetics, neurobiology, and behavior across a large group of individuals with autism and related conditions," says Dr. Schultz. "A partnership between the Center for Autism Research and the Seaver Autism Center brings us much closer to achieving this."

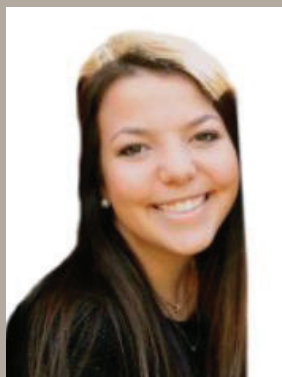


Interdisciplinary teams of CHOP and Seaver researchers work together to create collaborative project proposals.

Sinai Health System announced a formal clinical alliance in 2015, and this meeting between CHOP's Center for Autism Research and the Seaver Autism Center represents one of the first efforts towards a research collaboration between the two hospitals.

The day-long meeting started with a series of "Ignite Talks," in which clinicians and researchers from both teams presented future directions and opportunities in research across various disciplines,

INTRODUCING NEW TEAM MEMBERS



SAMANTHA BRIGHT

Samantha Bright joined the Seaver Autism Center as a Program Assistant in February 2017. Samantha graduated from Indiana University in May of 2016 with a Degree in Healthcare Management and Policy, and formerly interned at Mount Sinai in the Department of Neurosurgery.



LYNN HENDRICKSON, MBA

Lynn Hendrickson has taken on a new role at the Seaver Autism Center as the Administrative Director. Since she joined Mount Sinai in 2011, Lynn has supported the Department of Psychiatry as the Administrative Manager, and has also supported the Division of Psychiatric Genomics and the Seaver Autism Center. In her new role as Administrative Director of the Seaver Autism Center, Lynn will work with Seaver leadership to create and implement a strategic plan to guide the Center's growth in preclinical and clinical research, as well as expand the Center's outreach, education and training initiatives.

SEAVER STORY: JONAH

The Whitney family, upon discovering their son had a rare FOXP1 mutation, partnered with the Seaver Autism Center to help better understand FOXP1-related disorder.

As part of Autism Awareness Month, the Whitneys shared their #SeaverStory.

“Our son Jonah, is an energetic, engaging 12-year old with broad developmental challenges. After a long odyssey trying to understand the reason behind his developmental and behavioral challenges, visiting with many doctors and specialists along the way, we finally received a diagnosis when he was 8 years old. Whole-genome sequencing revealed that he has a point mutation in the FOXP1 gene. FOXP1 is a suspected autism-related gene.



As an infant, Jonah was very ‘colicky’ and had very low muscle tone. With physical therapy he sat on his own at 10 months and walked at 21 months. As he got older, it became clear that he had fine-motor and speech delays, as well as moderate intellectual disability. It also became clear that he had autistic-like features such as repetitive behaviors, narrow interests/obsessions, dislike of change or routine, and sensory issues. Other challenging behavior issues such as hyperactivity, impulsivity, inability to focus (ADHD) as well as anxiety also emerged and continue to pose our greatest challenges today. Although his social development is not ‘normal’ he is very engaged with people, so does not have a formal diagnosis of autism. We have struggled to find medications to help with his behavior issues. Jonah continues to progress at his

own pace and is doing well in school. He spends most of his time in a special needs class and gets support to participate in some mainstream classes. He continues to receive speech and occupational therapy at school. Outside of school, Jonah enjoys going to therapeutic horseriding lessons.

After Jonah’s diagnosis of a FOXP1 mutation, we were glad to finally have an answer to explain Jonah’s many issues, but were disappointed to discover that not much was known about FOXP1 in other patients. There were only a handful of cases reported in the literature, so we really didn’t have any more knowledge about Jonah’s prognosis. Since we both have backgrounds in science, we did as much reading as we could to learn about FOXP1. That is how we became aware of the researchers at the Seaver Autism Center. We also started an online community through RareConnect to find other families affected by FOXP1 gene issues. To date, over 30 families with affected children from around the world have joined the RareConnect FOXP1 community. This online community has provided an opportunity for families to share and learn from each other. A few families have been able to meet each other.

We knew that the Seaver Autism Center had experience in studying children with other single-gene changes and we connected with them to see if they could do the same for FOXP1. In March 2016, we visited with the team for an in-depth, extensive set of evaluations that have helped us understand Jonah’s current capabilities and additional strategies for helping him succeed in school and life. We remain actively engaged with the team at the Seaver Center, promoting their research to the wider FOXP1 community. Our shared goal is to characterize the syndrome, understand how it might change as the children develop, and most importantly to understand what techniques or therapies can help our children reach their highest potential. We are very grateful to the Seaver team for their efforts in advancing the understanding of FOXP1 so that the many families with affected children can have more knowledge and guidance to help them raise their unique children.”

– *The Whitney Family*



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Seaver Autism Center for Research and Treatment

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