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Seaver Researchers Featured at IMFAR 2015

At this year's International Meeting for Autism Research (IMFAR), sponsored by the International Society for Autism Research, work by over ten Seaver Autism Center researchers was featured in poster sessions, oral presentations, and in the annual press conference.

PRESS CONFERENCE

For the second consecutive year, research conducted by Seaver researchers led by Joseph Buxbaum, PhD, Director of the Seaver Autism Center, was featured in the annual IMFAR press conference. Of all the abstracts presented at IM-

FAR, the work led by Dr. Buxbaum was one of six presentations to be featured.

Dr. Buxbaum's presentation summary is reproduced below.

Phelan-McDermid syndrome is a genetic condition resulting in autism and intellectual disability. The syndrome is caused by mutation of the Shank3 gene, a gene which is important for brain function. Careful analysis of individuals with Phelan-McDermid syndrome has identified unique physiological and behavioral characteris-

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The Seaver Center also hosted our Third Annual IMFAR Reunion Reception for current and former members, friends, and collaborators.



As part of the Scientific Panel titled, "On the Road to Translational Treatments in Autism-Related Genetic Syndromes," Dr. Buxbaum presented, "Novel Findings in Phelan-McDermid Syndrome and their Translation into Therapeutics."

Seaver Associates Committee Listening Night

Earlier this spring, the Seaver Autism Center hosted its first ever "Listening Night" for the members of the Seaver Autism Center Associates Committee. This event was unique in that it involved the attendees speaking, and the scientific researchers listening. Seaver Center leadership had the important opportunity to see

through the eyes of parents and other relatives of people with autism, and to gain a new perspective. Participants enjoyed and appreciated this opportunity as well, and we look forward to hosting more Listening Nights in the future.

The Associates Committee is a group of dedicated stakeholders: parents,

grandparents, siblings, and others, who want to learn more and do more to support their loved ones with autism and to support the work being done at the Seaver Center. If you are interested in learning more, please contact Jessica Brownfeld (jessica.brownfeld@mssm.edu or 212-241-0349).

Autism Awareness Month at the Seaver Autism Center

This past April, the Seaver Center began several new initiatives for Autism Awareness Month, including the #SeaverStories social media campaign, a Mount Sinai mural, and weekly information tables.

SEAVER STORIES

In addition to the Fourth Annual Email Series for Autism Awareness Month, we executed the social media campaign #SeaverStories. Each week during the month of April, we featured an individual or family involved with the Seaver Center. Participants ranged from employees to patients and their families,



"Kali was involved in the IGF-1 trial through the Seaver Autism Center. We met so many great people and doctors, whom I felt had a genuine interest and concern for Kali. It was such an honor to be involved in this trial... We started with a little girl who couldn't walk, who would bite us because she was so frustrated, wouldn't look at us. We now have this beautiful little girl, who walks, doesn't bite, makes eye contact, laughs, giggles, gives hugs and kisses and is pretty much happy all the time!"
— Kraig and Krisha Kurtz

and through social media they shared their stories with the community. This campaign served as an opportunity to share the Seaver experience with a larger community. Those who followed this campaign were excited to learn more about the people involved and hear about their experiences with the Center.

To read the rest of these stories, visit us on Facebook at www.facebook.com/seaverautismcenter or search #SeaverStories on Twitter.

AROUND MOUNT SINAI



Members of the Seaver Autism Center team gather to commemorate Autism Awareness Month.



Marquette Hairston, Administrative Assistant in the Seaver Center, and Hillary Rieger, Clinical Research Coordinator, representing the Center at the first annual weekly informational tables for Autism Awareness Month.



First annual Mount Sinai wall mural with artwork by Jessica S. Johnson, MFA, Lab Manager for the Division of Psychiatric Genomics at Mount Sinai.

Director of Community Outreach Named to Museum Access Consortium Steering Committee

SEAVER AUTISM CENTER MUSEUM TOUR PROGRAM: THE DISCOVERY SQUAD

Established in 2013, the Seaver Autism Center has an ongoing collaboration with the American Museum of Natural History (AMNH), developed with Michelle Gorenstein-Holtzman, PsyD, Director of Community Outreach, and Danielle Halpern, PsyD, Director of Psychology Training. The Discovery Squad is a monthly tour program designed for children on the autism spectrum.

To learn more, visit <http://www.amnh.org/plan-your-visit/accessibility/autism-spectrum>.

MUSEUM ACCESS CONSORTIUM

As this experience was so rewarding, Dr. Gorenstein-Holtzman recently accepted a position to be on the steering committee for the Museum Access Consortium (MAC), which works with museums to help develop programs for individuals with disabilities.

Dr. Gorenstein-Holtzman is currently the only member of the committee representing a research institution, and this position will allow Dr. Gorenstein-Holtzman to bring more evidence-based practices into these programs.

For more information on the MAC, please visit <http://museu-maccessconsortium.org>.

tics, including specific areas of disability and strength. To better understand the neurobiology of the syndrome, we have introduced mutations in mouse and rat *Shank3* genes. The rat model is particularly useful for understanding the cellular basis of the disability in Phelan-McDermid syndrome. Rats have more complex social behavior, including juvenile play, and have brains that are more tractable to physiological analysis. We have observed that these rats have deficits in brain cell function, attention, and in social memory, and we are testing therapeutic approaches in these animals. Our recent results show that oxytocin, a safe and available treatment, has beneficial effects in the rat. Oxytocin reverses changes in brain cell function and enhances social memory. The implication

of these studies is that oxytocin may be a beneficial treatment in Phelan-McDermid syndrome, and we are approved by the US Food and Drug Administration to initiate such a trial.

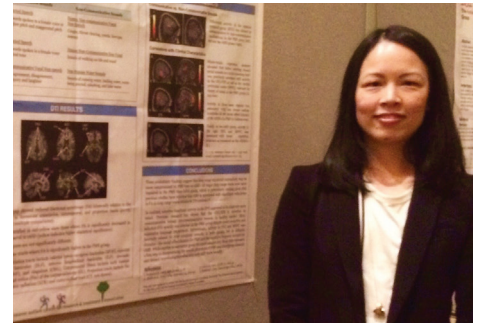
RESEARCH PRESENTATIONS

This year ten poster and oral presentations submitted by the Seaver Autism Center were accepted by IMFAR.

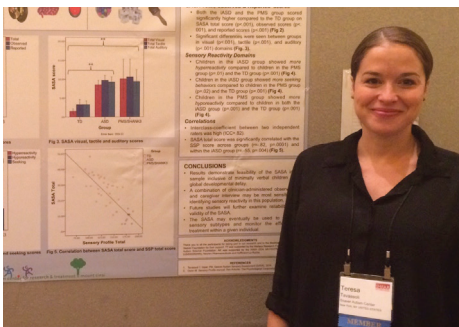
Hala Harony-Nicolas, PhD, Instructor in Psychiatry, gave an oral presentation titled, “Oxytocin Reverses Social Deficits in the *Shank3*-Deficient Rat, a First Genetically Modified Rat Model for Autism.”

Karim Ibrahim, MS, graduate student at the University of Hartford and former clinical psychology trainee at the Seaver Autism Center, gave a poster

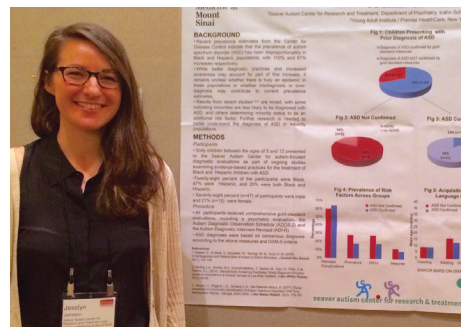
presentation titled, “Neural Predictors of Treatment Response to Social Skills Training in Children with Autism—Findings from a Randomized, Comparative Study.” Under the mentorship of Ting Wang, PhD, Mr. Ibrahim is conducting his graduate thesis research at the Seaver Center.



Ting Wang, PhD, Assistant Professor of Psychiatry, gave a poster presentation titled, “A Pilot Neuroimaging Study of Phelan-McDermid Syndrome.”



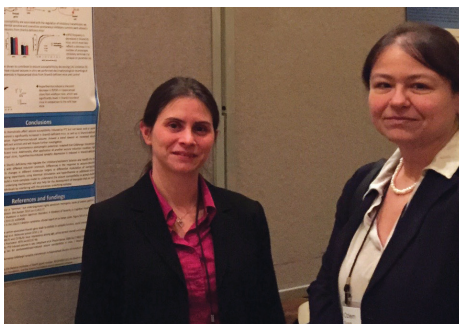
Teresa Tavassoli, PhD, Instructor in Psychiatry, gave a poster presentation titled, “SASA: A Sensory Reactivity Measure for Severely Affected Individuals with Global Developmental Delay and/or Autism Spectrum Disorder.”



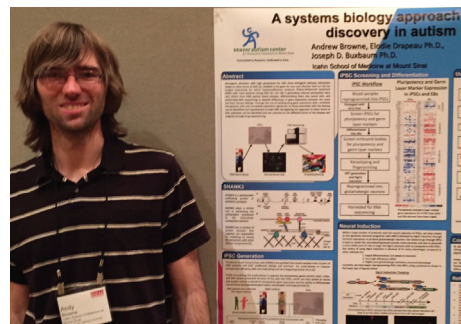
Jesslyn Jamison, BA, Clinical Research Coordinator, gave a poster presentation titled, “Accuracy of ASD Diagnoses in a Sample of Black and Hispanic School-Aged Children.”



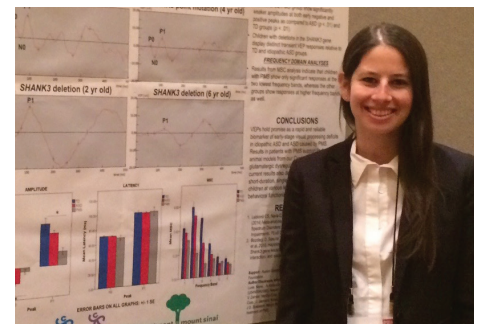
Silvia De Rubeis, PhD, Seaver Postdoctoral Fellow, gave an oral presentation titled, “Large-Scale Exome Analyses Reveal Novel ASD Genes Impacted By Genetic Variation at All Scales.”



Elodie Drapeau, PhD, Postdoctoral Fellow, and Ozlem Bozdagi Gunal, MD, PhD, Assistant Professor of Psychiatry, collaborated on a poster presentation titled, “Regulation of Seizure Susceptibility in *Shank3* Deficiency.”



Andrew Browne, MSc, Seaver Graduate Fellow, gave a poster presentation titled, “A Systems Biology Approach to Drug Discovery in Autism.”



Paige Siper, PhD, Instructor in Psychiatry, gave a poster presentation titled, “Transient Visual Evoked Potentials in Monogenic and Idiopathic ASD.”



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Mount Sinai Medical Center
One Gustave L. Levy Place, Box 1230, New York, NY 10029

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• **THE SEAVER AUTISM CENTER NEWSLETTER** brings you timely updates about new developments related to research and treatment of autism spectrum disorders, as well as activities at the Seaver Autism Center. To be placed on our mailing list, please contact SeaverCenterEditor@mssm.edu or The Seaver Autism Center, Icahn School of Medicine at Mount Sinai, One Gustave L. Levy Place, Box 1668, New York, NY 10029. Our phone number is 212.241.0961 and our website is www.SeaverAutismCenter.org.

• **SEAEVER IS CONTINUING TO GO GREEN!** Please send your email address to seavercentereditor@mssm.edu to receive this newsletter electronically.

Rare Disease Clinical Research Consortium Launch Meeting

This past spring the collaborators of the Rare Disease Clinical Research Consortium (RDCRC) titled, “Developmental Synaptopathies Associated with TSC, PTEN, and SHANK3 Mutations” met in Boston for a launch meeting. Funded by the National Institute of Neurological Disorders and Stroke, this Consortium will study the developmental disorders caused by mutations in three genes: TSC, PTEN and SHANK3.

This Consortium is one of over 20 consortia that make up the Rare Disease Clinical Network, an initiative of the Office of Rare Diseases Research.



Alex Kolevzon, MD, Clinical Director of the Seaver Autism Center, will lead the Phelan-McDermid syndrome longitudinal study as part of this RDCRC,

and Dr. Buxbaum will act as Administrative Director of the overall consortium. This grant marks the first time the National Institutes of Health has provided any funding for the clinical and longitudinal assessment of Phelan-McDermid Syndrome/SHANK3 mutations, which we now know to be a relatively common monogenic cause of autism. This study will comprehensively characterize the phenotype and natural history of PMS, identify biomarkers using neuroimaging, and identify genetic factors that contribute to diverse phenotypes in patients with PMS.



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DISTINGUISHED SCIENTIFIC SPEAKERS:
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AVI REICHENBERG, PhD
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