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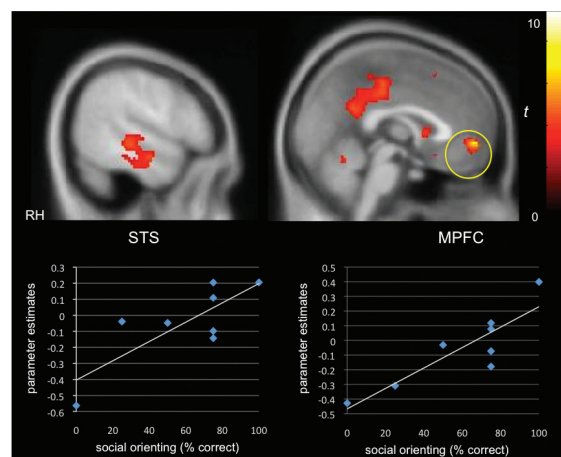
4 2016 SEAVER
AUTISM CENTER
CONFERENCE
SAVE THE DATE

New Research Published on Phelan-McDermid Syndrome

Seaver Autism Center researchers are the leading experts on Phelan-McDermid syndrome (PMS), a rare genetic syndrome which is associated with autism spectrum disorder (ASD). Researchers here have recently published two studies on this syndrome.

The first study appeared in the *Journal of Neurodevelopmental Disorders*, and it examined the differences in brain function between individuals affected by PMS and those with idiopathic ASD. The researchers found that children with PMS responded differently to communicative vocal sounds than children with idiopathic ASD, despite both groups possessing similar clinical characteristics.

The second study was published in the *Journal of Autism and Developmental Disorders*, and it compared sensory reactivity in children with PMS to children with idiopathic ASD. This study is the



In children with Phelan-McDermid syndrome, greater specialized “social brain” activity along the superior temporal sulcus (STS; left) and in the medial prefrontal cortex (MPFC; right) was associated with better orienting toward social sounds measured outside the scanner.

first to demonstrate differences in sensory reactivity between children with PMS and idiopathic ASD, helping to refine the PMS phenotype.

Collaborators Visit Mount Sinai

In April, the Seaver Autism Center hosted a visit for nurses in Sweden who work on the Population-based Autism Genetics and Environment Study (PAGES), funded by the National Institutes of Health. Led by Joseph D. Buxbaum, PhD, Director of the Seaver Autism Center, this study is a collaboration between the Seaver Autism Center at the Icahn School of Medicine at Mount Sinai, the University of Pittsburgh School of Medicine, Carnegie Mellon University, and the Karolinska Institutet (Sweden). The



L-R: Paige Siper, PhD (Chief Psychologist of the Seaver Autism Center), Anna-Carin Säll-Grahnat, Karin Dellenvall, Joseph Buxbaum, PhD, Christina Hultman, PhD, Birgitta Ohlander, Bozenna Iliadou, Ann-Britt Holmgren, Gun Karlsson, and Sven Sandin, PhD (Assistant Professor of Psychiatry at Mount Sinai).

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Collaborators Visit Mount Sinai

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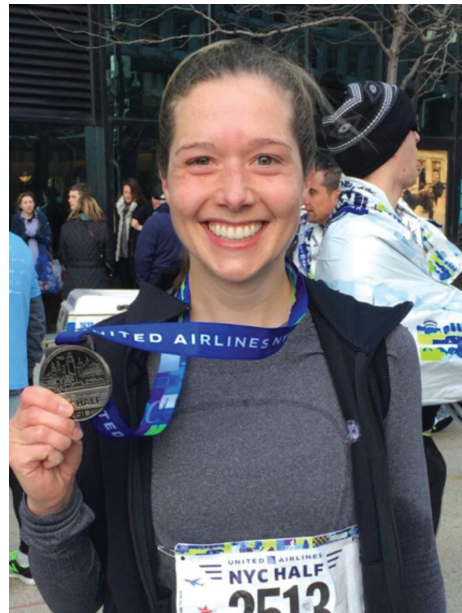
study uses a population-based epidemiological sample with detailed demographic and environmental information to assess the role of inherited and *de novo* genetic variants in autism. The researchers also evaluate rare standing variation in autism, while integrating key environmental variables.

As part of this study, this group of talented research nurses from the Karolinska Institutet, led by Professor Christina Hultman, PhD, collects hundreds of DNA samples every year. During the visit at Mount Sinai, the group participated in meetings to discuss psychiatric genetics with a focus on autism,

Obsessive-Compulsive Disorder and related disorders, and the preclinical and clinical research programs at the Seaver Autism Center. The group from the Karolinska Institutet and Seaver researchers alike enjoyed the opportunity to meet and discuss their work.

Clinical Fellow Runs for Seaver Autism Center

The Seaver Center thanks Lauren Donnelly, PhD, Seaver Clinical Fellow, for her fundraising efforts earlier this year. Dr. Donnelly ran the New York Half Marathon for the Seaver Center, and she raised \$3,400. We are very grateful for her time and generosity. Congratulations on an impressive performance, and thanks again to Dr. Donnelly!



Director of Community Outreach Named to Museum Advisory Board

Michelle Gorenstein-Holtzman, PsyD, Director of Community Outreach, was recently named to the Advisory Board of the Long Island Children's Museum. Dr. Gorenstein-Holtzman is also on the Steering Committee for the Museum Access Consortium.

Along with Danielle Halpern, PsyD, Director of Psychology Training, Dr. Gorenstein-Holtzman worked with the American Museum of Natural History to develop The Discovery Squad, a monthly tour program for children with autism. To book a tour, visit <http://www.amnh.org/plan-your-visit/accessibility/autism-spectrum/>

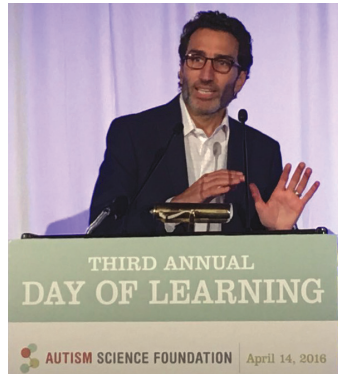
Recent Awards

- ▶ **2016 INTERNATIONAL MEETING FOR AUTISM RESEARCH (IMFAR) EARLY CAREER WORKSHOP TRAVEL AWARD**
Carla Golden, Graduate Student
- ▶ **KLINGENSTEIN THIRD GENERATION FELLOWSHIP AWARD IN ACCESS TO CARE**
Pilar Trelles, MD, Seaver Clinical Fellow
- ▶ **2016 IMFAR DIVERSITY AWARD**
Pilar Trelles, MD, Seaver Clinical Fellow
- ▶ **WINTER CONFERENCE ON BRAIN RESEARCH TRAVEL FELLOWSHIP**
Silvia De Rubeis, PhD, Seaver Postdoctoral Fellow

Highlights of Recent Presentations

▶ Joseph D. Buxbaum, PhD, recently gave a presentation titled “Combining Autism and Intellectual Disability Exome Data Yields Insight into both Disorders” at the American College of Neuropsychopharmacology Annual Meeting. Dr. Buxbaum also presented at the 6th Brain and Behavior Meeting of the Haifa Forum for Brain and Behavior (Haifa, Israel). His presentation was on “The Genetics of Autism: Insights into the molecular and cellular underpinnings of the social brain.”

▶ Alex Kolevzon, MD, Clinical Director of the Seaver Autism Center, presented at the recent Third Annual Day of Learning hosted by the Autism Science Foundation. His presentation was on “Understanding and Treating Anxiety in Autism.”



Alex Kolevzon, MD

▶ Julia George-Jones, Clinical Research Coordinator, gave a presentation titled “Electrophysiological Assessment of Idiopathic ASD” as part of Mount Sinai’s 18th Annual Child Health Research Day.

▶ Silvia De Rubeis, PhD, Instructor of Psychiatry, gave a presentation titled “Large-scale exome analyses reveal novel genetic variation in autism” at the 25th Neuropharmacology Conference.



Julia George-Jones

▶ Andrew Browne, Seaver Graduate Fellow, gave a presentation titled “Altered biological pathways in autism over the course of development” as part of the Systems and Developmental Biology and Birth Defects (SDBBD) course within the Graduate School of Biomedical Sciences at the Icahn School of Medicine at Mount Sinai.

Recent Events

▶ SPECIAL SEMINAR WITH JUNI PALMGREN, PHD

Juni Palmgren, PhD, Professor of Biostatistics at the Karolinska Institutet, recently presented a Special Seminar on “Research Infrastructure and Big Data.” Dr. Palmgren’s seminar emphasized the importance of “data intensive scientific discovery.”

▶ SEAVER SEMINAR SERIES

Jed Ellison, PhD, Assistant Professor in the Institute of Child Development at the University of Minnesota, recently visited Mount Sinai as part of the Seaver Seminar Series. His presentation was titled “Visual Orienting and Early Identification of Autism.”



Jed Ellison, PhD

Seaver Papers Selected by IACC for Top Advances in ASD Research

The Interagency Autism Coordinating Committee (IACC) recently released their 2014 and 2015 Summaries of Advances in Autism Spectrum Disorder Research. Each year, the IACC releases a list of “scientific advances that represent significant progress in the field.”

We are proud to announce that in these two years, researchers from the Seaver Autism Center were lead authors on five of the 40 papers. In addition, three additional papers by Seaver researchers were nominated.

SEAVER AUTISM CENTER PAPERS SELECTED IN 2014

▶ De Rubeis et al. Synaptic, transcriptional and chromatin genes disrupted in autism. *Nature*. 2014 Nov 13;515(7526):209-15.

▶ Pinto et al. Convergence of genes and cellular pathways dysregulated in autism spectrum disorders. *Am J Hum Genet*. 2014 May 1;94(5):677-94.

▶ Gaugler et al. Most genetic risk for autism resides with common

variation. *Nat Genet*. 2014 Aug;46(8):881-5.

▶ Sandin et al. The familial risk of autism. *JAMA*. 2014 May 7;311(17):1770-7.

SEAVER AUTISM CENTER PAPER SELECTED IN 2015

▶ Sanders et al. Insights into autism spectrum disorder genomic architecture and biology from 71 risk loci. *Neuron*. 2015 Sep 23;87(6):1215-33.



Mount Sinai

Seaver Autism Center for Research and Treatment

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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.

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



Mount Sinai *Seaver Autism Center for Research and Treatment*

SPECIAL

ALL TICKETS \$20
IN CELEBRATION OF OUR
20TH CONFERENCE
ANNIVERSARY

20TH ANNUAL
**Seaver Autism Center
Advances in Autism
Conference**

Save the Date
SUNDAY, SEPTEMBER 25, 2016

PRESENTED BY
THE SEAVER AUTISM
CENTER FOR RESEARCH
AND TREATMENT AT THE
ICAHN SCHOOL OF
MEDICINE AT MOUNT SINAI

NEW LOCATION
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COURSE DIRECTOR
JOSEPH D. BUXBAUM, PHD