seaver autism center
for Research & Treatment at Mount Sinai NEWS

Growth Hormone Clinical Trial

Phelan-McDermid syndrome (PMS) is caused by deletions or sequence variants of the SHANK3 gene and is a leading cause of autism spectrum disorder (ASD).

Insulin-Like Growth Factor-1 (IGF-1) is a hormone that promotes brain vessel growth, neurogenesis, and synaptogenesis. In early work at the Seaver Autism Center, IGF-1 reversed electrophysiological and behavioral deficits in Shank3-deficient mice (Bozdagi et al., 2013) and, critically, improved social withdrawal and restricted behaviors in children with PMS (Kolevzon et al., 2014). However, IGF-1 is difficult and costly to obtain and is associated with significant risks of hypoglycemia. Since IGF-1 can be increased innately by increasing growth hormone (GH) administration, this trial explored the use of GH in children with PMS and our pilot data demonstrate significant benefit across a wide range of clinical symptoms (Sethuram et al.,

2022). Based on these promising results, we have now started a larger, placebo-controlled clinical trial of GH in PMS and idiopathic autism (iASD).

The focus of this clinical trial is to use electrophysiological markers established in PMS to stratify subsets of individuals with iASD in order to predict treatment response. Our preliminary data demonstrate promising electrophysiological results using visual evoked potentials (VEPs). In addition, our preliminary work suggests some elements of the ASD phenotype in PMS that are unique, particularly as it relates to sensory reactivity abnormalities. Sensory symptoms are especially important because they are associated with VEP neural responses in both PMS and iASD populations, and they

may represent a phenotypic profile in PMS that informs which subset of individuals with iASD are most likely to benefit from intervention with GH.

To date, we have: 1) validated the ASD phenotype in PMS, 2) established electrophysiological biomarkers of PMS, 3) used biomarkers associated with PMS to identify an overlapping subset in iASD, and 4) begun a clinical trial with GH to use our biomarkers for stratification and to predict treatment responses. Upon successful completion of the trial, we expect to validate methods to predict treatment response based on electrophysiological profiles and to demonstrate the clinical efficacy of GH for individuals with PMS or iASD.

Increasing Diversity in the Field

The Seaver Autism Center has launched the Seaver Undergraduate Research Scholars Program.

This new training opportunity focuses on welcoming students from underrepresented groups in science and medicine (URiSM) through autism research.

Excellence and innovation in scientific research and education cannot be achieved without diversity, equity, and inclusion. Our team is committed to eliminating barriers and

increasing access to education and careers among students from (URiSM) and/or economically disadvantaged backgrounds.

Targeting these efforts toward trainees early in their career development will promote feasibility of entrance and encourage retention in science and medicine.

The Seaver Scholars Program will support the training and

mentoring of 14 outstanding undergraduate students in the 2022 calendar year. These students will be matched with a Seaver faculty mentor and hosted in their lab, where they will conduct innovative research aiming to better understand autism and develop new treatments.

Through this program, Scholars will have a unique opportunity to gain diverse skills, build confidence in interacting with scientific mentors, and boost

Increasing Diversity in the Field

their competitiveness when applying for subsequent academic positions.

We aim to support our Seaver Scholars by providing them with competitive stipends that allow them to choose research even if finances are tight.

You may sponsor a student or contribute any amount to help us reach our goal. Please, help us train the next class of scientific leaders in autism research and welcome much-needed diversity to our scientific ranks!

http://giving.mountsinai.org/goto/Seaver

Sensory Preferences

Not all sensory reactivity is the same. Visual hyporeactivity behavior is very different than tactile seeking behavior, and they should be treated and accommodated differently.

We are doubling-down on understanding sensory preferences, learning more about how to assess them, and using what we know to improve experiences. Our Chief Psychologist, Paige Siper, PhD is leading this breakthrough work.

Dr. Siper and her colleague Teresa Tavassoli, PhD, developed the *Sensory Assessment for Neurodevelopmental Disorders* (SAND). It is the only observational assessment and caregiver interview that classifies sensory reactivity for individuals of all ability levels.

SAND directly examines sensory hyperreactivity, hyporeactivity, and seeking behaviors across visual, tactile, and auditory modalities. This tool aids in treatment planning to target specific symptoms and helps to evaluate response to interventions.

SAND has been licensed by Stoelting and is available for purchase online.

COMING SOON:
Dr. Siper will be hosting clinical and research training classes. If you are interested in participating in the training, please fill out this brief form:
https://redcap.mountsinai.org/redcap/surveys/?s=TFXLLT3YNN

Improving the Patient Experience for Neurodiverse Individuals

It is common for people to feel uncomfortable when they go to a hospital or visit a doctor's office. There is harsh lighting, unusual sounds, and a hustle and bustle that can be unpleasant and even disruptive, especially for individuals on the spectrum. Because of this, vital medical attention is commonly delayed due to the stressors inherent in visiting a healthcare facility.

The Seaver team is working to improve the patient experience and transform care beyond our Center walls, across the health system. Our goal is to address the stressors that lead to delayed medical attention by ensuring that families with diverse needs will receive optimal care by clinicians who are properly trained and have tools to improve medical visits. To do so, we launched two exciting, new initiatives.

Sensory Kits

Our clinical team developed a Sensory Kit that can be distributed to patients in nearly any medical setting. The kit is a personal bag that the patient can keep with a few objects to satisfy sensory interests. The insert card describes how each item can be used to calm or stimulate a patient's nervous system to help improve their experience during their visit.

We were happy to call upon Spectrum Designs to help produce the kits. Spectrum Designs is a custom apparel and promotional products business with a social mission to create paid, meaningful, and inclusive employment opportunities for people with autism.

Staff Training

Just like everyone else, individuals on the spectrum have diverse medical needs that require professionals across the medical field to be trained on how to properly care for neurodiverse patients. Our team of psychologists developed the online psychoeducation training and assessment for Mount Sinai faculty and staff. The *Clinical Best Practices Educational Training for Treating Individuals with Autism and Related Conditions in a Medical Setting training* is currently being piloted at the Mount Sinai Beth Israel emergency department. After we receive feedback from the initial pilot, our goal is to roll out the training across the Mount Sinai Health System to ensure proper care for neurodiverse individuals is available in every department.

For more information about the Sensory Kits or the PEAK training, please contact Dr. Jessica Zweifach: jessica.zweifach@mssm.edu

National Institute of Mental Health F31/NRSA Fellowship

Congratulations to Seaver PhD Student, Amanda Leithead, for being selected for the prestigious NIMH F31/NRSA Fellowship! This fellowship is a training grant that is awarded to predoctoral students to fund their tuition/stipend.

Amanda's 3-year award supports her research that investigates pathways in the brain that are important for social behavior, and how genetic mutations associated with autism such as Shank3 might impact these pathways leading to deficits in social behavior.

Specifically, she will use mice to study the role of glutamate-oxytocin brain circuits in social behavior, and the effect of Shank3 mutations on these circuits.

In order to receive the NIMH F31/NRSA Fellowship Grant, she had to provide context about the collaborative training that she receives within the Seaver Autism Center and the Icahn School of Medicine at Mount Sinai. Being selected for the grant is a huge complement to Amanda and her mentor, Hala Harony-Nicolas, PhD, and also indicates the confidence NIMH has in our ability to provide premium training opportunities for our graduate students to advance the fields of science and medicine.

New Staff

ADAM SOCRATES, PHD

Adam completed his PhD at the Centre for Social, Genetic, and Developmental Psychiatry at King's College London and joined Dr. Eva Velthorst's lab as a post-doctoral fellow earlier this year. His work currently involves investigating the genetic factors that drive social withdrawal and the relationship between them and psychiatric disorders such as psychosis.

CHARIKLEIA CHATZIGEORGIOU, PHD

Charikleia joined Dr. Magda Janecka's team in the FunEpi lab in 2021. She completed her PhD at University of Leeds. At the University of Oxford, her previous research experience focused on identifying genetic risk factors for rheumatic autoimmune disease with the use of large-scale datasets. In the FunEpi lab, her focus is on the application of statistical genetics methods to unravel biological pathways of autism and other psychiatric disorders.

MIGUEL RODRIGUEZ DE LOS SANTOS, PHD

Miguel obtained his PhD at the Charité, University Medicine in Berlin, Germany in December 2021 and joined the Seaver Center in January as a postdoctoral fellow in Dr. Michael Breen's lab. He will investigate novel mutation-tailored therapeutic approaches for autism and rare genetic subtypes.

MARINA NATIVIDAD AVILA, MS

Marina is a Bioinformatician on the Seaver Center's Computational Genomics team. She attended the University of Glasgow for an MSc in Medical Genetics and Genomics and subsequently graduated from Boston University with an MS in Bioinformatics. She is working on the analysis of rare variation in populations with diverse ancestry.

PROMOTIONS!

Congratulations to the Seaver Team's newest Associate Professors

Decisions to promote faculty members are based on scientific productivity, funding support, and service to Mount Sinai and to the greater community.

We cannot be prouder of these amazing women in science on our team!

Silvia De Rubeis, PhD

Michelle Gorenstein, PsyD

Danielle Halpern, PsyD

Hala Harony-Nicolas, PhD

Jennifer Foss-Feig, PhD

Mount Sinai

Seaver Autism Center for Research and Treatment

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

[Facebook] [Instagram] @SeaverAutismCenter

[Twitter] @SeaverAutism

[Web] SeaverAutismCenter.org

• 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 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.

SAVE THE DATE

TUESDAY, MAY 24, 2022

Seaver Autism Center
26TH ANNUAL

Advances in Autism Conference

TOPIC
Convergence in Autism
From Many Genes to Few Pathways

HYBRID EVENT
Join online from anywhere or in-person at:
etc.venues
601 Lexington Avenue
New York, NY 10022

For more information please contact:
annualconference@seaverautismcenter.org

COURSE DIRECTOR
Joseph D. Buxbaum, PhD

Register now for FREE: https://bit.ly/advancesinautism22