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## Overcoming COVID Setbacks and Moving Research Forward

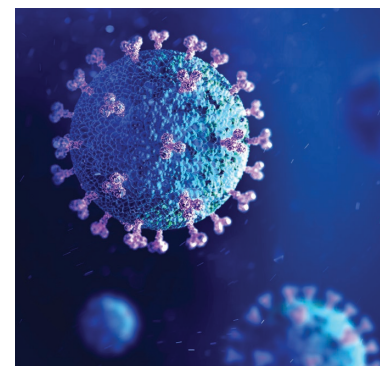
Navigating research projects during the COVID-19 pandemic has been the largest challenge at the Seaver Autism Center this year. In the heart of New York City, Mount Sinai Hospital found itself at the epicenter of the virus this spring. Like other institutions, we quickly established a plan to wind down our projects in order to prioritize the health of our patients, employees and research participants.

The Mount Sinai Health System quickly moved to an all-hands-on-deck effort to tackle the virus, both

on the frontlines and through new research initiatives. In response, the Seaver Center team utilized its existing expertise in patient-based research and genetics to initiate a study to collect biospecimens from individuals who tested positive for the virus with minimal symptoms, to compare with those collected from individuals with profound illness. Researchers are now using this information to identify the relationship of a person's genetic makeup and other biomarkers with the risk of developing severe

symptoms if infected, which can help identify best treatment practices and critical need for future vaccination.

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## The 24th Annual Advances in Autism Conference Went Virtual

On Thursday, September 17, the Seaver Autism Center hosted the 24th annual Advances in Autism Conference. Unlike years in the past, the event was presented entirely online. The virtual conference attracted a record number of attendees, bringing together over 900 participants, including academic experts in the field of

autism, healthcare professionals, parents and community groups.

This year, the focus of the conference was Profound Autism.

Autism is a spectrum, affecting each person diagnosed with the disorder in unique ways. Individuals who are more profoundly affected may have a lack of verbal and nonverbal communication skills, sensory challenges, cognitive difficulties, and often have simultaneous conditions such as epilepsy, anxiety and gastrointestinal diseases.

Our accomplished speakers presented material to help attendees further understand the needs of profoundly affected individuals.

Joseph D. Buxbaum, PhD, Director of the Seaver Autism Center, welcomed attendees to the day-

long event and began the program with a presentation that explained 'What We Know About the Causes of Autism and Its Comorbidities.'

Faculty members from the Seaver Center provided overviews of Sensory Reactivity in Neurodevelopmental Disorders (Paige Siper, PhD) and Novel Therapeutics in Autism – Bringing Industry and Academia Together (Ana Kostic, PhD).

We were also privileged to host a roster of exceptional guest speakers, all of whom are exceptional women and world experts from various institutions and disciplines. They presented the latest information about: Framework for Assessing Individuals With Rare Genetic Disorders Associated With

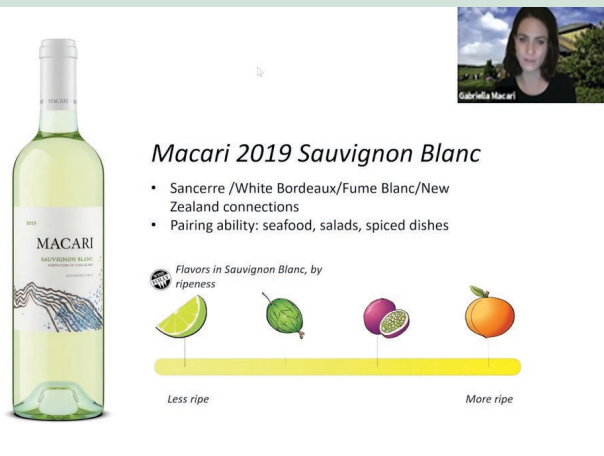
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## Sipping For Seaver

Amid the COVID-19 pandemic, we found a unique way to bring old and new Seaver Center supporters together for a safe, socially-distant, fun event – a virtual wine tasting.

The Sipping for Seaver event was hosted online on Thursday, October 15. Through a partnership with Macari Vineyards, we sold wine tasting packages that included three newly released wines from the vineyard - 2019 Sauvignon Blanc, 2019 Rose & 2015 Cabernet Franc – delivered directly to each participant's door ahead of the virtual tasting.



Certified Sommelier and Master of Wine candidate, Gabriella Macari, provided attendees with an entertaining and informative tasting, winemaking, and viticulture experience, along with a behind the scenes tour of the vineyard.

It has never been more challenging to keep autism research moving forward than this year. That is why we chose not to have a typical honoree during the virtual wine tasting and instead decided to recognize

our Teams at the Center. The Clinical, Pre-Clinical, Epidemiology, Drug Discovery and Development, and Administrative Teams have diligently continued to overcome challenges to ensure autism research remains a top priority.

The program included special remarks from the Director of the Seaver Autism Center, Joseph Buxbaum, PhD, that provided an update on the research landscape and progress that has been accomplished. Some of the highlights included teams returning to the lab and clinic, virtual clinical assessments, big data studies, the continuation of multiple for-benefit clinical trials, and new strategic partnerships to support our drug discovery and development efforts.

While our 2020 annual fundraiser looked different than we originally planned, we are thrilled that we were able to continue to recognize our Center's achievements and raise funds to push our research forward. The wine packages, sponsorships, and donations in honor of the celebration helped us raise over \$40,000 for the Center.

A special thank you to all of our sponsors and donors, and our Associates Board and Seaver Champions who helped contribute to the success of the event.

Please consider making a gift to help the Seaver Autism Center continue to advance autism research: [giving.mountsinai.org/SeaverCelebration](http://giving.mountsinai.org/SeaverCelebration)

## Overcoming COVID Setbacks and Moving Research Forward

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As New York City and Mount Sinai have reopened, members of our Center who are able to complete their jobs from home, such as those on the administrative and computational science teams, continue to work remotely to keep the density of the office spaces low. The basic science and clinical research teams stagger shifts while completing tasks remotely as possible.

To enable the Center's patient-based research to safely continue, the clinical team built programs to administer 80% of assessments remotely. When it becomes safe to bring in participants, they will only be required for a half-day in-person visit to complete the battery of tests, maximizing efficiently for the future.

Even through these difficult times, our teams continued to gain additional grant support and cultivate collaborative relationships for our Drug Discovery and Development Program. We are proud of the accomplishments our teams achieved in the face of adversity, and we continue to stay true to our mission to enhance the diagnosis of autism, discover its biological causes, and develop and disseminate breakthrough treatments.

We work to ensure autism research remains a priority - and we are eager to keep the needle moving forward to improve the lives of individuals and families affected by autism and related disorders.

## The 24th Annual Advances in Autism Conference

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Profound Intellectual and Multiple Disabilities (Audrey Thurm, PhD), Comorbidities and ASD: Distinct Disorders or Part of the Spectrum (Evdokia Anagnostou, MD), Communicating Signs of Distress (Eron Friedlaender, MD, MPH), Explorations of Language in Severe Autism (Helen Tager-Flusberg, PhD), and A Family's Journey to Treat Profound Autism (Amy S.F. Lutz).

We closed out the conference with a panel

of the day's speakers, moderated by Dr. Buxbaum, that answered questions submitted online from the audience throughout the day.

If you missed the conference, recordings of each presentation are available to watch online through April, 2021: <http://bit.ly/2020AdvancesinAutismPlayback>

Due to the extraordinary circumstances of this year, we enacted free admission to

the conference to ensure education was as accessible as possible during these trying times.

A special thank you to our event sponsors and donors - your support makes it possible for our Center to educate the community with the latest advances in research and care for individuals with autism.



# SEAVER CENTER HIGHLY CITED RESEARCHERS

Two leaders at the Seaver Autism Center have been recognized as Clarivate Analytics' 2020 'Web of Science' Highly Cited Researchers. Congratulations to Seaver Center Director, Joseph Buxbaum, PhD and Director of the Seaver Center's Epidemiology program, Avi Reichenberg, PhD!

Highly Cited Researchers have demonstrated significant and broad influence reflected in their publication of multiple papers that are highly cited by their peers over the course of the last decade. These highly cited papers rank in the top 1% of citations in the field.

## New Members of the Seaver Team



### MATT LALLI, PHD

Matthew joined the Seaver Center in September as an Instructor. He has a background in induced pluripotent stem cells (iPSC) disease modeling and functional genomics. He will develop and apply high-throughput approaches to study autism risk genes in human neurons.



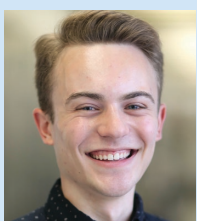
### SARAH BARKLEY

Sarah joined the Seaver Center as a Research Coordinator in August after graduating from Cornell University. As part of Dr. Jennifer Foss-Feig's group, she will be using neuroimaging and eye tracking methods to investigate biomarkers of idiopathic autism and related rare genetic disorders.



### SINJA (XUANJIA) FAN

Sinja graduated from Johns Hopkins University in May and returned to Dr. Michael Breen's group as an associate researcher. He is working on multiple studies involving blood RNA sequencing data and is also assisting in an RNA editing study.



### LIAM L. HIESTER

Liam is an associate researcher on the Drug Discovery and Development Team at the Seaver Autism Center. He studied at Rutgers University where he received his BA in Cell Biology and Neuroscience. His previous scientific training includes cell culture, immunofluorescence, confocal and fluorescent imaging, and quantification of neuronal survival and morphology. His current role is focused on identifying a phenotype for iPSC-derived neurons from Phelan-McDermid syndrome patients, and contributing to the identification of potential drug candidates for the treatment of autism.



### MICHAEL JUN

Michael is a native New Yorker with over 5 years of laboratory experience. He graduated from Brandeis University and worked in the Drug Discovery Lab at UCLA studying Alzheimer's and Parkinson's disease before joining the Seaver Autism Center's Drug Discovery and Development Team as an associate researcher. He is happy to be back in NYC with his family and working with a fantastic team.



### MICHELLE KIM

Michelle is a second year PhD student in neuroscience and joined Dr. Hala Harony-Nicolas' lab in April. She is interested in applying circuit dissection tools to study the development of the oxytocin system and its impact on social behavior in rodents. Prior to joining Mount Sinai, she worked as a research technician at UNC-Chapel Hill where she conducted research in ethanol consumption and feeding behavior in mice.



### BONNIE LERMAN

Bonnie joined the Seaver Center as a Research Coordinator in August, with a background in neuroscience and psychology from Duke University. She will be working on studies aimed to better characterize the phenotypes of various rare diseases, and investigating biomarkers of autism.



### ADELE MOSSA, PHD

Adele joined the Seaver Center in July as a Postdoctoral Research Fellow in Dr. Silvia De Rubeis' group. Her previous work focused on the molecular mechanisms underlying the pathophysiology of neurodevelopmental disorders using transgenic mouse models and pharmacological tools. At the Seaver Center, she will be working on the morphogenesis and synaptogenesis of cortical neurons in DDX3X syndrome. She will be also exploring the role of the cerebellum in DDX3X syndrome.



### YEAJI PARK

Yeaji joined the Seaver Autism Center in July after graduating from Macalester College with a degree in Neuroscience. She is working in Dr. Silvia De Rubeis' group, focused on understanding the mechanisms by which DDX3X syndrome affects neurodevelopment.



### REBECCA POLLAK, PHD

Becky joined the Seaver Center in August as a Postdoctoral Fellow working with Dr. Buxbaum's group. She earned her PhD at Emory University, where she studied rare genomic disorders associated with neurodevelopmental and psychiatric disease. Her work at the Seaver Center will focus on understanding the molecular and cellular mechanisms of DDX3X using human neuronal systems.



### MITZY RÍOS DE ANDA, PHD

Mitzy is a Postdoctoral Fellow on the Drug Discovery and Development Team at the Seaver Autism Center. Her background includes over five years of experience in stem cell, reprogramming, cancer, neurosciences and proteomics research. She received her PhD in Regenerative Medicine from the University of Edinburgh, where she described the proteomic molecular mechanisms that govern human stem cells and iPSCs. Her current research interests include the study of the most common single-gene causes of autism, such as Phelan-McDermid syndrome, for the discovery of potential drug candidates for its treatment.



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- **SEAVER IS CONTINUING TO GO GREEN!** Please send your email address to [seavercentereditor@mssm.edu](mailto:seavercentereditor@mssm.edu) to receive this newsletter electronically.

## DR. KOLEVZON RECOGNIZED AS RARE CHAMPION OF HOPE

Congratulations to the Seaver Autism Center's Clinical Director, Alex Kolevzon, MD, for being selected as a RARE Champion of Hope Awardee!

RARE Champions of Hope are groundbreakers and leaders who inspire and catalyze change in rare disease. This year, awardees were selected who have made a significant impact in advocacy, industry, medical care, and science, as well as up-and-coming rare disease leaders.

Each year, Global Genes, an organization dedicated to connect, empower, and inspire the rare disease community, opens nominations for the award to members of the rare disease community who know a Champion of Hope. All nominations are reviewed and honorees are selected by a Global Genes identified committee.

Dr. Kolevzon was nominated for the award by Ronni Blumenthal, Executive Director of the Phelan-McDermid Syndrome Foundation (PMSF).

"It's common knowledge that families who are lucky enough to bring their children to see Alex at the Seaver Center absolutely adore him," said Blumenthal. "But I'm not sure people know how much Alex does for the foundation behind the scenes."



Dr. Kolevzon chairs PMSF's Medical Advisory Committee, helped launch the foundation's ECHO program that connects local doctors with neurologists and psychologists who have expertise in Phelan-McDermid Syndrome (PMS), is a principal investigator for PMS research studies

and clinical trials, and is coordinating the development of PMSF's clinical consensus guidelines.

"In addition to all of his PMS projects, Alex always responds promptly to e-mails with questions or family concerns – even late at night or on the weekend," continued Blumenthal. "We are so grateful and fortunate to have him as a leader on our team."

Awardees were announced at this year's RARE Champion of Hope Celebration on November 12. The virtual event included exciting food and drink demos, entertaining performances, and inspiring stories of hope from the rare disease community.

We are immensely proud to have Dr. Kolevzon lead the clinical team at the Seaver Autism Center; his work has touched countless individuals and families impacted by autism and by rare neurodevelopmental disorders associated autism, including PMS, ADNP, DDX3X, and FOXP1 syndromes.