

## seaver autism center for Research & Treatment at Mount Sinai



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### A Seaver Clean Sweep - MCHDI 2021 Pilot Grants

The Mindich Child Health and Development Institute (MCHDI) awarded three pilot grants for the 2021 cycle. We are thrilled to announce that all of the projects that were selected have Principal Investigators (PIs) from the Seaver Autism Center!

These \$70,000 grants support early career scientists to pursue independently funded research projects. We are so proud of the talented women in science who are leading this exciting research! Learn more about the research projects:

#### **Probing Brain Circuitry Changes** in a Mouse Model of Autism and Intellectual Disability

Pls: Silvia De Rubeis, PhD, Seaver Autism Center & Zhuhao Wu, PhD Assistant Professor, Department of Cell, Developmental & Regenerative Biology & Department of Neuroscience

Mutations in the DDX3X gene are one of the most common causes of intellectual disability (ID) in girls and this gene has been identified as a high-risk gene for autism. Understanding the role of DDX3X in the formation of brain circuits will offer a new key to decipher ID and autism. This project will put together complementary expertise and innovative tools, applied to the

first mouse model with construct validity for DDX3X mutations. Based on findings in this new model, the team hypothesizes that DDX3X mutant mice have changes in neuronal populations that map onto specific circuits subserving brain function and behavior.

#### **Gene Expression in Endocervix During Pregnancy as a Novel Biomarkers of Neonatal Outcomes**

Pls: Magdalena Janecka, PhD, Seaver Autism Center, Lisa Eiland, MD, FAAP, Department of Pediatrics and Ernest Turro, PhD, Department of Genetics and Genomic Sciences

Neonatal adversity is associated with a host of short- and long-term health complications. Although these complications can be highly debilitating, and affect all body systems, our understanding of the mechanisms affecting neonatal adversity, and ability to predict it early in pregnancy, remain limited.

This project will address this knowledge gap by exploring the potential of using gene expression data from endocervical samples collected during routine examination of pregnant women. The team will measure gene expression of both coding and non-coding populations of RNAs. Then they will test the associations between gene expression - both

**CONTINUED** 

#### **Published!**

#### **PLOS Medicine:**

In the 'Gestational Age and the Risk of Autism Spectrum Disorder in Sweden, Finland, and Norway: A cohort study,' led by last author Sven Sandin, Associate Professor at the Seaver Autism Center, it was observed that the relative risk of autism increased weekly as the date of delivery diverged from 40 weeks, both pre- and- post term. The overall risk of autism was low, especially for girls born after 42 weeks of gestation, but the risk increased for each week of gestational age before or after 40 weeks.

Learn More:

http://bit.ly/AgeAndRiskofAutism

#### Frontiers in Psychiatry:

A study led by first author Jen Foss-Feig, PhD, Assistant Professor at the Seaver Autism Center looked at whether EEG could be used to predict later onset of psychosis in adolescents and young adults with autism showing early clinical warning signs. The team found that it could, but in an opposite way than is typically seen in non-autism samples: in autism, stronger, rather than weaker, EEG response was associated with later onset of psychotic illness. The study is important because it suggests that EEG could contribute to more accurate predictions about disease trajectory for psychosis, and that knowing about a person's autism diagnosis may be important for accurately interpreting EEG findings in this context.

Learn More:

http://bit.ly/ConversionToPsychosis

# A Seaver Clean Sweep – MCHDI 2021 Pilot Grants

#### CONTINUED

at a single RNA, and pathway levels – and subsequent birth outcomes (including gestational age and size for gestational age; APGAR score; NICU admission).

#### A Family-Mediated Intervention to Improve Outcomes in Minority Children with Autism Spectrum Disorder affected by the COVID-19 Pandemic

**Pls:** Pilar Trelles, MD and Jennifer Foss-Feig, PhD, Seaver Autism Center

Social distancing measures introduced to mitigate the spread of coronavirus have dramatically impacted the utility of treatment programs for children with autism. As a consequence, there has been a dramatic increase in need for psychiatric services and hospitalizations. In minority groups, these effects are further exacerbated by existing racial and ethnic healthcare disparities.

As a consequence of the COVID-19 pandemic, the Center Ann Sullivan of Peru (CASP) developed a 16-week, virtual, parent-mediated curriculum designed to support families during this period, building on an evidence-based program dating back to 1984. The intervention empowers caregivers to help their children build on functional skills considered essential to promote independence, facilitate academic learning, and enable inclusion in all aspects of life. The grant will enable Drs. Trelles and Foss-Feig to evaluate the utility of the CASP online curriculum, compared to treatment as usual, in underserved minority youth with autism in New York City.

## Personal Fundraisers to Support Research at The Seaver Autism Center

Birthday fundraisers and sponsored runs are just a few ideas to rally friends and family to help support a good cause. Did you know Mount Sinai has a platform to create online fundraising pages to make giving back even easier? You can create a webpage customized with your story and a photo or video to share with family and friends to help raise money for the research done at the Seaver Autism Center.

Simply visit: **giving.mountsinai.org/getinvolved** and select your fundraiser type from the options listed.



**Get Started!** Select to join as a new participant. Fill out the questionnaire and you'll be directed to your personal participant center. In your participant center, you can customize your page and create a unique URL to share easily through your social media pages, emails, or text messages.

Select 'Edit Personal Page' from the menu on the right side to start personalizing. Share your story by adding a title and the reason you are raising money for the Seaver Center.



The work that we do at our Center is not possible without the help of supporters like you! If you are interested in creating a fundraiser or have any questions when setting up your fundraising page, email Whitney Cortes, who can help guide you through the process: Whitney.Cortes@mountsinai.org

### Dr. De Rubeis, Distinguished Scholar



Congratulations to Dr. Silvia De Rubeis for being selected as a recipient of the second annual Distinguished Scholar Award from the Icahn School of Medicine at Mount Sinai, sponsored by the Office of Gender Equity in Science and Medicine.

Dr. De Rubeis' project "Decoding Sex-Specific Drivers of Intellectual Disability" aims to understand the mechanisms underlying sex differences in ID by focusing on the risk gene DDX3X.

Loss of one of two copies of DDX3X in females causes DDX3X Syndrome associated with autism

and intellectual disability, while males only have one copy. Why does cognitive development require two functional DDX3X copies in females but not in males? This project seeks to address this question by studying how DDX3X regulates the molecular and cellular development of the cerebral cortex in female and male mice.

The award is intended to help Assistant Professors with independent research programs maintain their momentum as they integrate family caretaking responsibilities into their careers.

#### New Class of Seaver Fellows

The Beatrice and Samuel A. Seaver Foundation extended two previous fellowships and approved five new fellowships for 2021. The Seaver Fellowship Program enhances all areas of research at the Seaver Autism Center. Year after year, it has proven to be a successful platform to help launch careers for young scientists. We are grateful to the Seaver Foundation for their ongoing support to enhance the field of autism research.

Learn more about the new fellowship projects:

#### POSTDOCTORAL FELLOW



ADELE MOSSA, PhD Mentor: Silvia De Rubeis, PhD

Dr. Mossa will study the role of the cerebellum brain region in DDX3X syndrome. The cerebellum is important for both motor and social function, and patients with DDX3X have deficits in both. She will use behavioral, cellular, and molecular techniques to investigate how the cerebellum develops and functions in DDX3X mutant mice. The project will provide an understanding of how

the development of the brain is altered in DDX3X syndrome.

#### POSTDOCTORAL FELLOW



CHRISTINE HANSEN, PhD Mentor: Magdalena Janecka, PhD

It has been hypothesized that one of the key biological processes in autism is dysregulation of on/off switching of different genes, resulting in the brain receiving too much or too little of the chemicals that typically guide its development. Dr. Hansen's research focuses on identifying epigenetic events (called "epivariations") that are rare, but have large impacts on gene on/off switching, and

assess how damaging they are likely to be by inspecting if they are also present in unaffected family members.

#### **FACULTY SCHOLAR**



#### MATTHEW LALLI, PhD Mentor: Joseph Buxbaum, PhD

Dr. Lalli is an Instructor at the Seaver Center. His work focuses on large-scale studies to get from the many autism genes to a few pathways. He will develop and apply high-throughput approaches to study autism risk genes in human neurons. This work involves disrupting top autism genes in cell models and measuring RNA and protein, in order to map genes to a much smaller number of

pathways. This will in turn provide insights into major molecular pathways for interventions, whether or not an individual carries an autism mutation.

#### POSTDOCTORAL FELLOW



#### WEIYAO YIN, PhD Mentor: Sven Sandin, PhD

Dr. Yin's research project will perform the largest and most detailed examination of cancer risk in children with autism to date. She will use one of the most recognized sources for cancer research provided by the Swedish cancer register, which records all malignant tumors in Sweden since 1958 and collaborate with some of the world's most recognized experts in cancer epidemiology.

#### **GRADUATE STUDENT**



#### SARAH BANKER

Mentor: Jen Foss-Feig, PhD

Sarah will use state-of-the-art tools to image the brains of individuals with autism and neurotypical individuals while they perform virtual reality-like tasks that measure the ability to engage in simulated social interactions. Using mathematical modeling, she will explore how various brain regions function differently in relation to social processes in autism vs. controls and

contribute to social symptoms in autism. The results of this project will offer new tools for identifying categories of autism and provide novel insight into treatment.

#### **NEW SEAVER TEAM MEMBERS**



#### RYN CUDDLESTON, MSc

Ryn graduated in May 2020 with their MSc from the University of Nevada Reno. As a PhD student through the genetics and data science program, Ryn joined the Breen lab in February 2021, where their work will explore RNA editing in the developing brain and in neurological disorders.



#### CHRISTINE HANSEN, PhD

Christine has a PhD from the University of Copenhagen in bioinformatics and biostatistics with a focus on epigenetics of psychiatric disorders. In the Janecka lab, her focus is on epigenetic mechanisms of prenatal development leading towards adult morbidity in psychiatry leveraging family cohorts.





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- 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 18, 2021** 

Seaver Autism Center 25TH ANNUAL

## **Advances in Autism Conference**

**TOPIC** 

# **Quantifying Autism**Increasing Feasibility of Clinical Trials

**VIRTUAL CONFERENCE** 

**COURSE DIRECTOR** 

Access link will be distributed following registration

Joseph D. Buxbaum, PhD

For more information please contact: annual conference@seaverautismcenter.org

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