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FOXP1 Syndrome added to GeneReviews

Thanks to a valuable contribution from members Dr. Siper, Dr. Kolevzon, and Dr. Kostic of the Seaver Autism Center, FOXP1 syndrome is now included in GeneReviews. GeneReviews is an important database describing specific genetic diseases. Each entry includes information about the genetic basis of the disorder, diagnostic criteria, possible treatments for various symptoms, and family support services.

FOXP1 syndrome is one of several rare genetic syndromes included in the Seaver Autism Center's Rare Disease Program. FOXP1 syndrome is a neurodevelopmental disorder caused by disruptions to the Forkhead Box Protein P1 (FOXP1) gene. The Seaver Center has shown that mutations in FOXP1 have a significantly identified cause of autism. Common

features include autism, (including significant sensory symptoms and repetitive behaviors), mild-to-severe intellectual disability, language impairment, attention deficits, behavioral challenges and a host of medical conditions. Since 2017, The Seaver Autism Center has published a number of critical studies describing the clinical phenotype of FOXP1 syndrome, including practice parameters for medical assessment and monitoring. Recently, our group completed the first study examining post-pubertal psychiatric and medical changes in FOXP1 syndrome. As described by Dr. Siper, "Preliminary results are positive and suggest continued development of skills even at later ages."

In parallel to observational studies in individuals with

FOXP1 syndrome, the Center continues its preclinical research to better understand the mechanistic underpinnings of neurodevelopmental changes observed in the clinic. There are also a number of collaborations focused on leveraging induced pluripotent stem cell (iPSC) lines derived from patients and their family members to develop preclinical assays and test potential drug candidates.

FOXP1 syndrome's inclusion in GeneReviews is a major step forward – establishing a reference that will promote greater awareness of the disorder as well as a more comprehensive understanding of the knowledge and tools currently at our disposal, thereby improving treatment.

The Guggenheim Museum's *Guggenheim for All*

The Seaver Autism Center's Chief Psychologist, Dr. Paige Siper, recently completed a study in collaboration with the Guggenheim Museum's Guggenheim for All (GFA) program. Results from the study are now published in the international peer-reviewed journal, *Journal of Museum Education*. The Guggenheim for All (GFA) program was developed to increase access to high quality arts programming for neurodiverse individuals. The study, designed by Dr. Siper, examined the efficacy of a manualized arts educator training developed by Melanie Adsit (co-lead author). The training offers arts educators evidence-based recommended practices to incorporate into art lessons with neurodiverse audiences.

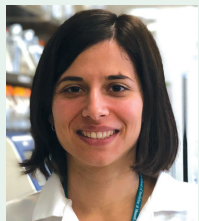
The paper, *Guggenheim for All: Virtual Arts Programming for Autistic Students and the Efficacy of a Manualized Training for Arts Educators*, demonstrated (1) significantly improved educator fidelity in implementing evidence-based practices in virtual art classes for autistic individuals, and (2) significantly increased parent-

reported sensitivity of the educator to their child's individual needs, following the training. De-identified videos of virtual arts classes were collected prior to and following educator training and coded by blinded raters who did not know which classes were conducted pre- or post-training. Seaver Autism Center clinical research coordinators were the raters for this study.

Dr. Siper described the impact of this initiative: "This work has the potential to change the landscape of arts accessibility by opening doors to arts programming led by trained educators. Educators, who are uniquely skilled to ensure people of all abilities can participate in exceptional arts experiences." Furthermore, the virtual training and education model allows for widespread dissemination across other cultural institutions. To learn more about the Guggenheim For All program described in this article, visit www.guggenheim.org/accessibility/guggenheim-for-all/guggenheim-for-all-toolkit.

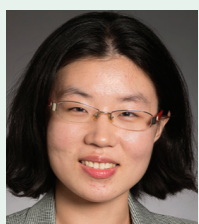
Young Investigator Grants from the Brain & Behavior Research Foundation

Two Seaver Team members were recently awarded Young Investigator grants from the Brain & Behavior Research Foundation. BBRF Young Investigator Grants provide each awardee with up to \$35,000 per year for two years (\$70,000 total) to enable promising young scientists to continue research fellowship training or to begin careers as independent researchers. The mission of the program is to support researchers in launching careers in neuroscience and psychiatry.



Marta Garcia-Forn, PhD, joined the Seaver Autism Center as a postdoc in the De Rubeis lab in January 2020. With her Young Investigator Grant, she aims to study the role of the *DDX3X* gene in the generation and function of glutamatergic brain cells, and its contribution to anxiety-like behaviors. Mutations in the X-linked RNA helicase *DDX3X*

cause a neurodevelopmental disorder affecting primarily females and manifesting with intellectual disability, autism spectrum disorder, motor dysfunction and anxiety. The project is expected to uncover a better understanding of how cortical brain development shapes brain cell diversity that contributes to complex behaviors. Ultimately, the project should allow researchers to begin to understand the comorbidity between autism and other psychiatric conditions.



Xuran Wang, PhD, joined the Seaver Autism Center as an Assistant Professor in August 2022. Xuran plans to develop a statistical framework that integrates single-cell and tissue-level RNA transcriptomes with genetic variation to better understand how alterations in cell composition and cell-type specific expression contribute to the

etiology of psychiatric disorders. By identifying the genetic mechanisms that underlie changes in cell composition and gene expression, she aims to contribute to the development of new therapeutic targets for mental illnesses with this research. Her project will focus on developing machine learning models to uncover the biological mechanisms that contribute to mental illnesses: by identifying the genetic and cellular factors that contribute to psychiatric disorders, she plans to gain a better understanding of how these conditions develop, and to identify new treatment options.

Uplifting Athletes Foundation Young Investigator Draft Grant

Marta Garcia-Forn has been awarded a \$20,000 grant, sponsored equally by the DDX3X Foundation and the Uplifting Athletes Foundation.



The Young Investigator Draft is the result of Uplifting Athletes' ongoing commitment to accelerate scientific advancements for rare disease treatments by supporting the next generation of researchers. The organization is represented by chapters sponsored by over 30 athletic teams across the country. Representation by prominent athletes has encouraged awareness of rare diseases, and has strengthened a sense of community among patients, families, advocates, researchers, and allies.

On February 3, Marta was presented with her award and matched with an athlete at the Draft Night ceremony at the Lincoln Financial Field, the home of the Philadelphia Eagles: "The Draft Night was a fantastic event... it was inspiring and emotional to hear how the athletes' stories are connected to rare diseases and how they're doing a great job of raising awareness about them," said Marta. "The night was a reminder of how important it is to keep doing research on rare diseases."



Marta's athlete match is Cooper Dawson, football player for Charleston Southern University. Cooper Dawson is the founder and President of the Uplifting Athletes chapter at his university. He is motivated to use his platform to spread awareness about rare diseases, sharing: "My friend Kingsley, who has cerebral palsy, inspires me. I learned that the only disability is a bad attitude."

Marta will be using the grant to study *DDX3X* syndrome, described above. She plans to use a mouse model to study the congenital brain malformations in cortical brain development that occur in those affected by the syndrome. Marta stated, "The intricacies of brain development captivate me, and receiving this award enables me to persist in studying it within the context of *DDX3X* syndrome. My aim is to uncover new therapeutic targets for those affected."

MCHDI Pilot Program Grants

Seaver Team member Dr. De Rubeis and colleague Dr. Martin-Trujillo have received the prestigious Mindich Child Health and Development Institute Pilot Project grant. Projects focus on advancing knowledge of child health through collaborative efforts. The study has been funded with \$75,000 for the period between March 1, 2024 and March 1, 2025.



Dr. De Rubeis and Dr. Martin-Trujillo will integrate genomics and neuroscience approaches to study Tandem Repeat Expansions (TREs) associated with neurodevelopmental disorders. TREs represent a class of genetic mutations that are emerging as a significant source of risk for human disorders. The scientists will present their work at the Mindich Child Health and Development Institute's retreat in November 2024.

New Staff



Tahshin Rahman

Tahshin Rahman joined Seaver Autism Center in October 2023 as a Grants Manager. With years of experience in Federal and Non-Federal Grants, Tahshin excels in identifying, securing, and managing grants to support impactful projects.



Catherine Sancimino, PsyD

Dr. Sancimino is a Clinical Psychologist who joined the Seaver Autism Center as an Assistant Professor. She has provided clinical evaluations for research at Albert Einstein College of Medicine, NYU, and Rutgers University. Her previous experience includes assessment and intervention for autistic children and adults as well as for individuals who have experienced traumatic events. Dr. Sancimino has provided psychological services in schools, hospitals, and community mental health clinics. Her long-term goals include contributing to effective treatments for profound autism through clinical research as well as supporting children and their families by providing assessment and intervention.



Shiraz Bheda, MSc

Shiraz is a biostatistician whose primary focus is the application of statistical methods to better understand the relationship between underlying genetic data and disease. Prior to joining Seaver, he was a member of the Center for Precision Medicine & Genomics (CPMG) at Columbia University, and before that, Shiraz worked as a quantitative financial analyst at Caxton Associates, a macroeconomic-oriented hedge fund. Shiraz has a Master of Science degree in Statistics and Statistical Analysis from Stevens Institute of Technology.



Shlomit Beker, PhD

Dr. Beker is a cognitive neuroscientist, focusing on studying physiological mechanisms of cognitive disorders. She joined the Seaver faculty following her post-doctoral training at Albert Einstein College of Medicine, where she studied altered cognitive functions in children and adults with autism. Her research seeks to identify physiological measures of altered synchronization between individuals with autism, and their physical and social environment, using EEG, behavior and measures from the body. Alongside the goal of elucidating the understanding of the latter, Dr. Beker's long-term goal is to use these noninvasive readouts to advance biomarkers for diagnosis and treatment of the disorder.



Corinne Smith

Corinne, a first-year Neuroscience PhD student, recently joined Silvia De Rubeis's lab. She graduated from Florida State University with a degree in Psychology, supplemented by extensive coursework in chemistry and biology. Prior to pursuing her doctoral studies, Corinne worked as a research technician at Weill Cornell, focusing on understanding the impact of a transcription factor on brain development. Driven by her passion for developmental neuroscience and a keen interest in neurodevelopmental disorders, Corinne is eager to apply her expertise to the research endeavors at the Seaver Center.

RNA Club Founded at Mount Sinai



Seaver Team Faculty member Michael Breen, PhD, has recently established an innovative RNA Club at Mount Sinai. Ribonucleic acid (RNA) is a crucial molecule responsible for translating the DNA instructions of the genome into functional proteins within cells. Dr. Breen's research at the Seaver Center is centered on RNA editing as well as the genetics and genomics of rare neurodevelopmental disorders.

Designed to foster a collaborative environment, the RNA Club aims to cut across multiple departments and research domains, inviting researchers, clinicians, and students within the hospital system to participate in an exchange of the latest research findings. This dialogue is facilitated through internal meetings and special sessions featuring distinguished external speakers, held on a monthly basis, ensuring a continuous dialogue within the field.

Dr. Breen is particularly enthusiastic about the collaborative nature of the RNA Club, stating, "The Club was born out of our love for RNA biology, recognizing its importance across multiple domains of human health, diseases, and therapeutics. The idea is to create a more holistic consensus of RNA experts with different but complementary sets of skills."

RNA research has the potential to illuminate new pathways for understanding and treating autism: "Our collaborative approach is

not just about advancing our knowledge of RNA, but about making meaningful strides in comprehending and addressing the intricacies of autism, potentially opening new avenues for therapy and intervention." RNA therapies are a rapidly growing area for behavioral health.





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