

# SATB2 RESEARCH ROUNDTABLE SPEAKERS

JUNE 22<sup>ND</sup> 2023



# Dr. Nico Wahl

Institute for Neuroscience, Georg Dechant lab, Medical University of Innsbruck, Austria

*My background is in molecular biology with a focus on next generation sequencing approaches. We study the underlying mechanisms of neuropsychiatric disorders by investigating how genes are regulated in the cortex. The aim of our research is to understand the pathways involved in consolidating cognitive functions. We have recently shown that SATB2 organizes the genome in the brain specifically at genic regions important for cognition.*



# Dr. Andrew G. Newman

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Institute of Cell Biology and Neurobiology,  
Charité – Universitätsmedizin Berlin, Germany

*Our lab focuses on studying the genetic basis of cerebral cortex development. We are interested in identifying genes whose disruption causes brain malformation and investigating molecular functions of proteins encoded by such genes.*



# Paulina Żamejć

Mother of Łukasz, diagnosed with SATB2-Associated Syndrome, Poland

*I am a mother of two, living in Wrocław in Poland. My son Łukasz is a young adult with SATB2-Associated Syndrome. He is my coach and mentor; he never gives up and that's what he has been teaching me every day for the past 16 years. Through the years there has been many challenges, medical tests, behavioural and communication issues, but also small and big successes in his development that we achieved with persistence and strong will.*

*I have been my son's advocate since he was born, but in the past year I have started also a new role as a patient advocate on a national level, being one of the drivers of the SAS community in Poland.*



# Prof. Bin Chen

Molecular, Cell & Departmental Biology  
University of California, Santa Cruz, USA

*My background is in the field of molecular and developmental neuroscience. My laboratory investigates neural stem cell lineage and neuronal cell fate specification in the developing cerebral cortex. We have found that during development, multi-potent neural stem cells sequentially generate diverse neuronal cell types in the cerebral cortex, followed by producing cortical glial cell types. We have identified multiple transcription factors that are important for different cortical neuronal subtypes to establish their identities. We are currently studying how these transcription factors regulating gene expression in the cortical neurons.*



# Dr. Joery den Hoed

Language and Genetics Department,  
Max Planck Institute for Psycholinguistics, the Netherlands

*As a molecular biologist, I am particularly interested in how we can use (high-throughput) assays using human cells to learn more about rare genetic variants disrupting brain development - confirming their pathogenicity, exploring potential disease mechanisms, but also establishing genotype-phenotype correlations. During my doctoral studies, I worked on neurodevelopmental disorders associated with the SATB1 gene. Now, as a postdoctoral researcher in the research group of Prof. Simon Fisher, I extended these studies to investigate SATB2 as well.*



# D.V.M, Dr. Shinya Yamamoto

Department of Molecular and Human Genetics, Baylor College of Medicine, USA

*Our lab studies many rare and undiagnosed diseases using the fruit fly (*Drosophila melanogaster*) in collaboration with clinicians around the world. We function as part of the Model Organisms Screening Center of the Undiagnosed Diseases Network, an NIH-funded large clinical consortium. We also serve as the *Drosophila* Core of the Center for Precision Medicine Modeling at Baylor College of Medicine, an initiative to support clinical research using the power of genetic model organisms.*

*We started to work on SATB2 about 1.5 years ago because we were highly inspired by parents of a SAS patient who carried a novel variant in this gene. We recently established an *in vivo* assay in flies to classify SAS variants into different functional classes, which we believe will have a broad clinical and translational impact.*



# Dr. David Bright

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SATB2 Gene Foundation, Board Member/Research Lead  
Department of Pharmaceutical Sciences, Ferris State University  
College of Pharmacy, Michigan, USA

*I am a proud parent of Lydia, age 10, who was diagnosed with SATB2-Associated Syndrome in 2017. I serve as a Board Member and Research Lead for the SATB2 Gene Foundation. I am also a Professor of Pharmacy where my research focuses on clinical service implementation and improvement, most recently with clinical pharmacogenomics.*





# Dr. Kathryn Post DeMott

Odylia Therapeutics, Scientist, USA

*I am a neuroscientist with a background studying neurodegenerative as well as neurodevelopmental disorders. I have always had a passion for the rare disease community since growing up with a cousin who has a rare neurodevelopmental disease.*

*Through my work at Odylia I help develop gene therapies for rare diseases as well as partner with patient advocacy groups to assist in their therapeutic development goals.*



# Dr. Yuri Zarate

Chief, Division of Genetics and Metabolism,  
University of Kentucky, USA

*I am a clinical geneticist and I'm interested in craniofacial disorders. I lead the only SAS multidisciplinary clinic in the US and have evaluated dozens of patients with SAS. I have participated in different research SAS-related efforts for the last several years and helped delineate in greater detail the phenotype.*



# Dr. Jennifer Fish

Department of Biological Sciences,  
University of Massachusetts Lowell, USA

*My lab uses diverse experimental systems and techniques to investigate molecular, cellular, and developmental mechanisms underlying craniofacial and limb development. My background includes a PhD dissertation in the cell biology of neurogenesis, and post-doctoral training in craniofacial development and skeletal biology at King's College London and the University of California, San Francisco.*

*In my current role as a principal investigator, I have become very interested in variation in penetrance and severity of congenital anomalies. A major focus of my research for the past 15 years has been to understand mechanisms underlying morphological variation in craniofacial morphogenesis associated with a reduction in dosage of the important developmental genes, *Fgf8* and *Satb2*. *Fgf8* is a signaling factor expressed primarily in the epithelia of the developing pharyngeal region, while *Satb2* is a chromatin modifier and transcription factor expressed predominantly in the neural crest of the pharyngeal arches. Craniofacial development exhibits dosage-dependent responses to both of these genes. We aim to elucidate molecular mechanisms mediating these dosage responses during tissue development.*



# Dalal Dawood Baumgartner

Founder of SATB2 Connect and mother of Naomi, diagnosed with SATB2-Associated Syndrome, Australia.

*Dalal is the founder and principal director of SATB2 Connect, a charity operating in all aspects of the organisation's four pillars; Support, Research, Advocacy & Networking; ensuring all individuals and families receive the optimal opportunity for better outcomes, where none are left behind. Combining her passion for project management, teaching and rare advocacy, she is determined to drive research opportunities and collaborate with local universities and organisations and international SATB2 organisation partners.*

*Together with:*

**Rachel Muir**

Educational genetic counselor, Australia

**Carolina Barbero & Siwon Kang & Wesley Zhang**

Students at University of Sydney, School of BioMedical Engineering, Australia



# Dr. Joe Zhou

Division of Regenerative Medicine, Department of Medicine, Weill Medical College of Cornell University

Director of Human Therapeutic Organoid Core at Weill Cornell Medicine, USA

*Dr. Zhou received his PhD training in neuroscience at the California Institute of Technology. He then focused on studies of pancreatic islet regeneration first as a postdoctoral fellow, then as a faculty member at Harvard University where he became Associate Professor before moving to Weill Cornell Medicine. His laboratory in New York studies pancreatic islet and intestinal mucosa regeneration. Pertinent to SAS, his lab investigates the role of SATB2 in controlling colonic mucosal biology relevant to diseases including SAS and inflammatory bowel diseases.*



*Participates in the discussion panel:*

# Dr. Luigi Boccuto

School of Nursing, CBSHS, Healthcare Genetics Interdisciplinary  
Doctoral Program, Clemson, USA

*My background is in clinical genetics. My work at the School of Nursing lab (Clemson University) is focused on the characterization of genotype-phenotype correlations in genetic disorders. We study metabolic profiles of cells from individuals with genetic conditions in order to investigate metabolic pathways involved in pathogenic mechanisms, identify potential biomarkers, identify molecular targets for treatment approaches, and assess the efficacy and side effects of candidate drugs.*

*We are interested in collaborating with clinical centers to collect information on phenotypes, biospecimens for functional studies, and eventually develop novel treatment approaches.*

