

INSYNC-AS

INTERNATIONAL ANGELMAN
SYNDROME RESEARCH
COUNCIL

SPONSORED BY



SFARI SIMONS FOUNDATION
AUTISM RESEARCH INITIATIVE

INSYNC-AS 2022 Meeting | JUNE 9-10 | New York

Location: Simons Foundation: 160 Fifth Avenue (Entrance on 21st Street)
Gerald D. Fischbach Auditorium, 2nd Floor

Zoom: [https://simonsfoundation.zoom.us/j/95153627462?](https://simonsfoundation.zoom.us/j/95153627462?pwd=OUswMktyaGFxa2Q4VUJEdVU4YWYyZz09)
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Meeting ID: 951 5362 7462

Passcode: 236606

▸ Welcome Letter

▸ Mission

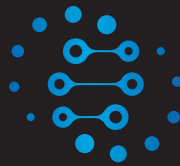
▸ Agenda

▸ INSYNC Council Members

▸ Speakers



WELCOME



Dear Guests and INSYNC Council members,

Welcome to this 2nd annual meeting of the International Angelman Syndrome Research Council (INSYNC-AS).

We are grateful that you have agreed to participate in this effort to build a community of scientists, clinicians, pharmaceutical executives, regulators, and other thought leaders, to accelerate drug development for Angelman syndrome and other related neurodevelopmental disorders (NDDs).

Our hope is that these discussions will help to inform funding strategies that build on the recent successes in Angelman syndrome, will encourage research in areas where gaps need to be filled, and will help support how these learnings can be applied to other NDDs.

This year we are focusing Day 1 on the current state of drug development in Angelman syndrome and bringing in key experts on how to best improve diagnostic and patient identification efforts globally as more gene therapy and disease modifying therapies are reaching the clinics. We will then end the day with a focus on biomarkers being developed for Angelman syndrome through the Angelman syndrome Biomarker and Outcome Measure Consortium (ABOM).

On Day 2 we will be focusing on 2 other NDDs, Rett syndrome and STXBP1 Disorders. Our team of council members and invited speakers will work to support key opinion leaders for these disorders and support their drug development efforts meeting them where they are in their journey.

We are excited about the progress in this field, but also realize that these are still early days. By bringing diverse talent together we hope to encourage and accelerate the highest quality work to bring meaningful change to the lives of all individuals living with NDDs.

Thank you for being a part of this effort!

Allyson Berent, DVM, DACVIM

Chief Science Officer, Foundation for Angelman Syndrome Therapeutics
Co-Director, INSYNC-AS
Co-Director, Angelman Syndrome Biomarker and Outcome Measure Consortium (ABOM)
Chief Operating Officer, GeneTx Biotherapeutics
Director of Interventional Endoscopy, Internal Medicine, The Animal Medical Center
Mother, Quincy 8 years old (deletion + AS)
allyson.berent@cureangelman.org

John E. Spiro, PhD

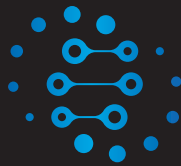
Deputy Scientific Director, Simons Foundation Autism Research Initiative (SFARI)
Co-Director, INSYNC-AS
jspiro@simonsfoundation.org

Jennifer Panagoulas, RAC

Co-Director, INSYNC-AS
Regulatory Advisor, Foundation for Angelman Syndrome Therapeutics
Co-Director, Angelman Syndrome Biomarker and Outcome Measure Consortium (ABOM)
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MISSION



Mission

INSYNC-AS: A collaboration between FAST and the Simons Foundation Autism Research Initiative (SFARI)

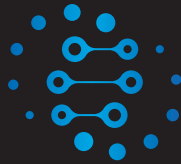
We, at the Foundation for Angelman Syndrome Therapeutics, are continuing to demonstrate our commitment to identifying and furthering scientific advancements for various potential therapeutics for Angelman syndrome (AS) through an exciting collaboration with the Simons Foundation Autism Research Initiative (SFARI). INternational Angelman SYNdrome Research Council (INSYNC-AS) actively evaluates and drives research initiatives in Angelman syndrome, and other similar neurodevelopmental disorders (NDDs), using a robust integrated approach of tapping into the expertise and experience from the foremost thought leaders in diverse fields. This Council not only includes experts focusing on Angelman syndrome currently, but also aims to bring in key opinion leaders working in many other arenas in order to further grow the translational research roadmap for AS.

THE GOALS OF INSYNC-AS

The goal of INSYNC-AS is to build a community of collaborative advisors by leveraging the combined skill sets of scientists, clinicians, geneticists, pharmaceutical executives, and other key opinion leaders in order to empower further drug development for Angelman syndrome. This Council of Excellence, or “Brain Trust,” will provide innovative ideas, ensure all potential research avenues are identified, explore novel therapeutic platforms, and further de-risk those platforms which are heavily invested. INSYNC will also identify gaps in current AS research and encourage new initiatives to fill them. The Council of Excellence will highlight, a multi-functional combination of skill sets and expertise to help prioritize FAST’s deep funding strategies, while also leveraging these learnings to other NDDs.

Another goal of this consortium is to encourage strong and clear consensus amongst pre-clinical and clinical colleagues working in neurodevelopmental disorders (NDD) of the best way to clinically test novel therapeutic compounds to ensure neurobehavioral testing is consistent, of the highest quality, and integrating the latest strategy in regulatory practice. These goals will help FAST’s mission, to ensure all patients living with AS will see meaningful and transformative therapeutic benefits, regardless of age or genotype, and through that apply as many learnings as possible to many other NDDs.

AGENDA



INSYNC-AS 2022 Meeting

Thursday June 9, 2022 | 9:00am – 5:00pm EDT, Cocktails and Dinner to follow

Friday June 10, 2022 | 8:30am – 3:00pm EDT

Simons Foundation: 160 Fifth Avenue (Entrance on 21st Street)

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Agenda – Day One (June 9): Angelman Syndrome

8:00 – 9:00am	Breakfast
9:00 – 9:10am	Welcome Allyson Berent DVM (FAST); John Spiro PhD (SFARI); Jennifer Panagoulis, RAC (FAST)
9:10 – 10:25am	Update and Overview on Drug Development in AS
9:10 – 10:00am	Allyson Berent DVM (FAST) <i>Landscape for Drug Development in Angelman Syndrome: Annual Update and New Initiatives</i>
10:00 – 10:25am	Discussion on Landscape/Patient Identification Initiative- FAST Global Search & Rescue moderated by Jennifer Panagoulis, RAC (FAST)
10:25 – 10:40am	Coffee Break
10:40 – 11:30am	The Diagnostic Odyssey
10:40 – 11:05am	Lynne Bird, MD (Rady's Childrens Hospital) <i>The Diagnostic Odyssey in Angelman Syndrome: Clinician's Perspective</i>
11:05 – 11:15am	Alana Newhouse (FAST) <i>Why a Diagnosis Matters: A Parent's Perspective</i>
11:15 – 11:30am	Discussion on the Best Way to Improve The Diagnostic Journey moderated by Laurent Servais, MD, PhD (Oxford University) and Elliott Sherr, MD, PhD (University of California, San Francisco)
11:30am – 2:10pm	Improving Patient Identification Efforts: FAST Global Search and Rescue Initiative
11:30am – 12:00pm	Anne Wheeler, PhD (RTI) <i>Incidence and Prevalence Challenges in AS: ICD-10, Claims Database, NBS, Literature Review and Available Data</i>



12:00 – 12:15pm	Laurent Servais, MD, PhD (Oxford University) <i>How Patient Identification Was Accelerated in Other Rare Diseases: SMA/DMD/Other</i>
12:15 – 12:45pm	Discussion on Creating an Infrastructure to Identify Patients Globally: Undiagnosed, Misdiagnosed, Not Connected moderated by Wendy Chung, MD, PhD (SFARI)
12:45 – 1:30pm	Lunch
1:30 – 1:50pm	Yael Weiss, MD, PhD (Mahzi Therapeutics) <i>How to Leverage the Expertise of Diagnostic Vendors to Support Patient Identification for a Community and Industry</i>
1:50 – 2:10pm	Discussion on Leveraging For-Profit Companies to Support Patient Identification and Community moderated by Omar Khwaja, MD, PhD (VectivBio)
2:10 – 3:15pm	Data Collection Best Practices for FAST Global Search and Rescue
2:10 – 2:25pm	Jennifer Panagoulas, RAC (FAST) <i>How to Collect Patient Identification Data that is Most Effective and Useful for Community and Industry</i>
2:25 – 2:55pm	Anne Wheeler, PhD (RTI) Honey Heussler, MD (Queensland Hospital) Meagan Cross (FAST-AU) Matthew Bellgard, PhD (Queensland University) <i>Leveraging Existing Databases: GASR and LADDER</i>
2:55 – 3:15pm	Discussion on Database Best Practices and Creating an Infrastructure Globally moderated by Jennifer Panagoulas, RAC (FAST)
3:15 – 3:30pm	Coffee Break
3:30 – 5:30pm	Angelman Syndrome Biomarker and Outcome Measure Consortium (ABOM)
3:30 – 4:15pm	Allyson Berent, DVM (FAST); Joerg Hipp, PhD (Roche) <i>Introduction to Novel Endpoints and Pharmacodynamic Biomarkers for AS</i>
4:15 – 4:25pm	John Foxe, MS, PhD (University of Rochester) <i>Translational Neurophysiological Markers in Neurodevelopmental Disorders</i>
4:25 – 5:00pm	Discussion on PD Biomarkers moderated by Rachael Hawtin, PhD (Ultragenyx)
5:00 – 5:30pm	Open Session to Create Action Steps for FAST Global S&R
5:30 – 6:30pm	Cocktail Reception on Promenade
6:45 – 8:00pm	Dinner at Blackburn (19 E 26th St)



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Agenda – Day Two (June 10): Rett Syndrome and STXBP1 Disorders

7:30 – 8:30am Breakfast

Welcome

8:30 – 9:00am **Rett Syndrome CURE 360 Overview**

8:30 – 8:40am **Jana von Hehn, PhD** (Rett Syndrome Research Trust)
Strategy to Cure Rett Syndrome

8:40 – 8:50am **Randall Carpenter, MD** (Rett Syndrome Research Trust; Allos Pharma)
Biomarker Consortium, Pipeline

8:50 – 9:00am Discussion moderated by **Yael Weiss, MD, PhD** (Mahzi Therapeutics)

9:00 – 9:30am **Molecular Biomarkers**

9:00 – 9:20am **Victor Faundez, MD, PhD** (Emory University School of Medicine)
Rett Syndrome Biomarkers: A Search among the Secreted Proteomes of the Brain and its Cells

9:20 – 9:30am Discussion moderated by **Ashley Winslow, PhD** (Odylia Therapeutics)

9:30 – 10:00am **EEG Biomarker**

9:30 – 9:50am **Joerg Hipp, PhD** (Roche)
EEG Biomarkers in a Rare Genetic Neurodevelopmental Disorder

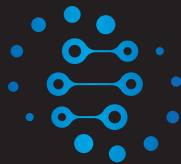
9:50 – 10:00am Discussion moderated by **Paul Wang, MD** (SFARI)

10:00 – 10:10am Break

10:10 – 11:30am	Digital Biomarkers
10:10 – 10:30am	Dudley Tabakin, MSc (VivoSense) <i>Wearable Biosensors</i>
10:30 – 10:50am	Dina Katabi, MS, PhD (Massachusetts Institute of Technology) <i>Invisible Biosensors</i>
10:50 – 11:10am	Gari Clifford, DPhil (Emory University and Georgia Institute of Technology) <i>Combining Wearables and Nearables for Tracking Changes in Health</i>
11:10 – 11:30am	Discussion moderated by John Spiro, PhD (SFARI)
11:30 – 11:45am	STXBP1 Disorders Introductory Remarks
	Charlene Son Rigby, MBA (RARE-X; STXBP1 Foundation)
11:45 – 12:05pm	What is Known about STXBP1 Disorders Biology
11:45 – 11:55am	James Goss, PhD (STXBP1 Foundation) <i>Molecular Mechanisms, Cellular and Animal Models</i>
11:55 – 12:05pm	Discussion moderated by Yong-Hui Jiang, MD, PhD (Yale University)
12:05 – 12:45pm	Lunch
12:45 – 1:25pm	Clinical Landscape for STXBP1 Disorders
12:45 – 1:15pm	Ingo Helbig, MD (Children's Hospital of Philadelphia) <i>Natural History and Disease Concept Model</i>
1:15 – 1:25pm	Discussion moderated by Zachary Grinspan, MD, MS (Weill Cornell Medicine)
1:25 – 2:05pm	Translational Research Landscape for STXBP1 Disorders
1:25 – 1:40pm	Ganna Balagura, MD, PhD (Vrije Universiteit Amsterdam) <i>ESCO: an EU Consortium to Promote Trial Readiness for STXBP1 Disorders</i>
1:40 – 1:55pm	Matthijs Verhage, PhD (Vrije Universiteit & Amsterdam University Medical Center) <i>Connecting Cellular Phenotypes, qEEG and Clinical Symptoms</i>

1:55 – 2:05pm	Discussion moderated by Elliott Sherr, MD, PhD (University of California, San Francisco) & Joerg Hipp, PhD (Roche)
2:05 – 2:45pm	Current State of the Drug Development Pipeline for STXBP1 Disorders
2:05 – 2:25pm	Michael Boland, PhD (Columbia University Irving Medical Center) <i>An Overview of Gene Targeted Strategies and Drug Discovery Platforms to Treat STXBP1 Haploinsufficiency</i>
2:25 – 2:45pm	Discussion moderated by Stuart Cobb, PhD (University of Edinburgh; Neurogene Inc.)
2:45 – 3:00 pm	Closing Remarks
	John Spiro, PhD (SFARI) Allyson Berent, DVM, DACVIM (Foundation for Angelman Syndrome Therapeutics; GeneTx Biotherapeutics) Jennifer Panagoulas, RAC (Foundation for Angelman Syndrome Therapeutics, Angelman Syndrome Biomarker and Outcome Measure Consortium) Monica Coenraads, MBA (Rett Syndrome Research Trust) Charlene Son Rigby, MBA (RARE-X; STXBP1 Foundation)

INSYNC COUNCIL MEMBERS



**Barbara Bailus, PhD****Keck Graduate Institute, Claremont Colleges**

Dr. Barbara Bailus is an Assistant Professor of Genetics at The Keck Graduate Institute, of the Claremont Colleges. She did her doctoral research at University of California, Davis in the Segal Laboratory developing a gene therapy based on artificial transcription factors for the treatment of Angelman syndrome. Her postdoctoral research was in the Ellerby Lab at The Buck Institute for Research on Aging, focused on potential therapies for Huntington's disease including small molecules and gene editing proteins. In 2021 Dr. Bailus was honored to take on the role of Chair of the Scientific Advisory Board for the Foundation for Angelman Syndrome Therapeutics (FAST). Previously, Dr. Bailus served as a scientific advisor and communications reporter for FAST. Dr. Bailus has a passion for teaching the next generation of scientists and communicating exciting scientific advances to the public. When not in the lab or classroom she enjoys being outside, traveling, riding horses, and painting.



Arthur Beaudet, MD
Cofounder and CEO Luna Genetics, Inc.

Dr. Arthur Beaudet received his MD from Yale University, did his pediatric residency at Johns Hopkins, and was a research associate at the NIH before joining Baylor College of Medicine (BCM) in 1971 where he remained until 2020. Dr. Beaudet has made diverse contributions in the field of mammalian genetics including discovery of uniparental disomy in humans. In 1994, his lab first reported that the UBE3A gene mapped in the Angelman region, in 1997 he co-discovered that mutations in the gene cause Angelman, and in 2015, in collaboration with Isis Pharmaceuticals (now Ionis), Dr. Beaudet showed that oligonucleotides could be used to activate the paternal copy of the gene in the mouse as a therapeutic strategy. In 1995, Dr. Beaudet was elected to the National Academy of Medicine and in 2011, he was elected to the National Academy of Sciences. Dr. Beaudet is currently the cofounder and CEO of Luna Genetics, which is focused on launching cell-based noninvasive prenatal testing (NIPT) as a new form of genetic prenatal diagnosis.


Allyson Berent, DVM, DACVIM
Foundation for Angelman Syndrome Therapeutics; GeneTx Biotherapeutics
Interventional Endoscopy Services, Animal Medical Center, NYC
Co-Director Angelman Syndrome Biomarker and Outcome Measure Consortium

Dr. Allyson Berent is a veterinary internal medicine specialist who serves as the Director of Interventional Endoscopy Services at the largest animal hospital in the world, The Animal Medical Center, in New York City. After graduating from Cornell University College of Veterinary Medicine she completed an internship at the University of Minnesota and a residency in Small Animal Internal Medicine at the Veterinary Hospital of the University of Pennsylvania. After completing a fellowship in interventional radiology at the Veterinary Hospital of the University of Pennsylvania, a fellowship in Endourology at Thomas Jefferson University, and an Interventional radiology fellowship at the Hospital of the University of Pennsylvania, she served as an Adjunct Assistant Professor in Internal Medicine and Interventional Radiology/ Interventional Endoscopy at the Matthew J. Ryan Veterinary Hospital of the University of Pennsylvania. Dr. Berent has a particular focus on medical device development, stem cell therapy through regenerative medicine and selective arterial delivery, ureteral diseases, urinary incontinence and minimally invasive management of upper tract urinary obstructions and biliary obstructions. In 2014, Dr. Berent's daughter was diagnosed with a rare non-degenerative neurogenetic disorder called Angelman syndrome. In October of 2015 she joined to Board of Directors as a Scientific Director for the Foundation for Angelman Syndrome Therapeutics (FAST), and in March of 2016, became the Chief Science Officer for the Foundation. Dr. Berent helped to spearhead the development of a pre-competitive biomarker and outcome measure consortium in order to bring patient focused outcome measures forward for human clinical trials (Angelman Syndrome Biomarker and Outcome Measure Consortium-ABOM) and Co-Founded the International Angelman Syndrome Research Council (INSYNC). Through FAST, Dr. Berent collaborated with a consortium of scientists to encourage translational research opportunities, in order to help bring novel genetic therapies forward toward human clinical trials. Through this work, with the foundation, Dr. Berent co-founded GeneTx Biotherapeutics, a company singularly focused to advance an antisense oligonucleotide (ASO) therapy through IND enabling studies and a phase 1/2 clinical trial. Dr. Berent currently serves as the Chief Operating Officer of GeneTx Biotherapeutics, who partnered with Ultragenyx Pharmaceuticals in August of 2019. The Phase 1/2 clinical trial for the safety and tolerability of GTX-102 started enrolling patients in February 2020 as the first intrathecally delivered ASO for Angelman syndrome.



Elizabeth Berry-Kravis, MD, PhD
Rush University Medical Center

Elizabeth Berry-Kravis MD, PhD is a Professor of Pediatrics, Neurological Sciences, and Biochemistry at Rush University Medical Center in Chicago. In 1991, she established the Fragile X Clinic and Research Program, through which she provides care to over 700 patients with fragile X syndrome (FXS). She has studied medical issues, epilepsy and psychopharmacology in FXS, and has been a leader in translational research in FXS including development of outcome measures and biomarkers, natural history studies, newborn screening, and particularly clinical trials of new targeted treatments in FXS. Her laboratory studies the cellular role of fragile X mental retardation protein (FMRP), relationship between FMRP and clinical function, and optimization of genetic testing methods. More recently, Dr. Berry-Kravis has expanded clinical and translational work to other neurodevelopmental disorders and genetic neurodegenerative diseases including autism spectrum disorders, Phelan McDermid syndrome, Rett syndrome, Angelman syndrome, Niemann-Pick type C, Batten's disease, pantothenate kinase-associated neurodegeneration, and creatine transporter deficiency.

**Stuart Cobb, PhD****Patrick Wild Centre and Simons Initiative for the Developing Brain, University of Edinburgh; Neurogene**

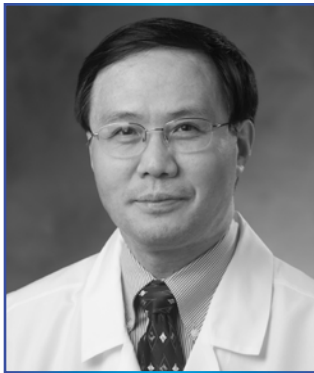
Dr. Stuart Cobb heads a translational research laboratory that is focused on developing genetic therapies for severe neurological and neurodevelopmental disorders. His research aims to address the tractability of severe brain disease to genetic rescue and to develop innovative therapeutic solutions for clinical translation. His research highlights include the original ground-breaking genetic rescue results in Rett syndrome, and his group was the first to report the ameliorative effect of gene therapy in mice modelling the disorder. His laboratory has developed a number of novel gene therapy approaches optimized for efficacy and safety. This includes the development of regulated expression cassettes, minigene and RNA based approaches. Dr. Cobb was a founding member of the RSRT Gene Therapy Consortium. Since late 2018 he has been Chief Scientific Officer at Neurogene, a genetic therapy company focusing on rare neurological disease.



Joseph Gleeson, MD
University of California, San Diego

Dr. Joseph Gleeson is the Rady Professor of Neurosciences and Pediatrics at UCSD, Director of Neurosciences at the Rady Children's Institute for Genomic Medicine, and CMO of the n-Lorem Foundation. His goal is to understand causes of pediatric brain diseases including autism, epilepsy and spina bifida, to improve outcomes. The lab has sequenced a cohort of 10,000 patients, and uncovered over hundreds of causes of disease, including recessive

and somatic mutations, and determined that many conditions have potential points of treatment. The n-Lorem Foundation seeks to develop and provide personalized antisense drugs to patients 'for free, for life'. Dr. Gleeson received an MD degree from the University of Chicago, residency in pediatrics and neurology at Children's Hospital, Boston and research fellowship at Harvard Medical School. Awards include Simons Foundation Autism Research Initiative Investigator, Howard Hughes Investigator, Klingenstein Award in the Neurosciences, the Searle Scholars Award, the Burroughs Wellcome Fund Translational Research Award, and the 2017 Lieber Prize for Innovation in Developmental Neuroscience. Dr. Gleeson was named a member of the National Academy of Medicine in 2013.



Yong-Hui Jiang, MD, PhD
Yale University

Dr. Yong-Hui Jiang is a Professor of Genetics, Neuroscience, and Pediatrics at the Yale University School of Medicine and Chief of Medical Genetics at Yale Medicine and Yale New Haven Health.

Dr. Jiang is a physician, scientist and active practicing physician at Yale Children's Hospital and Yale New Haven Hospital. He received his MD at Shanghai Medical College of Fudan University in China and his PhD in Human and Molecular Genetics at Baylor

College of Medicine under the mentorship of Dr Arthur Beaudet. Dr. Jiang completed the residency in pediatrics and fellowship in clinical genetics and medical biochemical genetics at the Texas Children's Hospital and Baylor College of Medicine. His clinical interests are genetic evaluation of neurodevelopmental disorders, inborn error of metabolisms, and rare disease genomics. Dr. Jiang's research program focuses on investigations of genetic and neurological bases of human neurodevelopmental disorders including autism in humans using new genomic technologies and in animal models using interdisciplinary approach. Special interests include investigation of Angelman and Prader-Willi syndromes and SHANK3, SHANK2, SCN2A, UBE3A, and HISTH1E genes causing autism and neurodevelopmental disorders.



Omar Khwaja, MD, PhD
VectivBio AG

Dr. Omar Khwaja is the Chief Medical Officer of VectivBio AG, a global biotechnology company focused on the discovery and development of transformative treatments for severe rare conditions with high unmet medical need. Prior to VectivBio, Dr. Khwaja was Chief Medical Officer and Head of Research and Development at Voyager Therapeutics, a neuroscience gene therapy company in Boston, MA. He began his career in drug development at Roche, leading programs in neurodevelopmental disorders, where he was Head of Rare Diseases as well as Neuroscience Early Development. A practicing pediatric neurologist and geneticist, Dr. Khwaja received his MD and PhD from the University of Cambridge. He trained in pediatrics, genetics and neurology in London, Melbourne and Boston, where he became Chief Resident at Boston Children's Hospital. Dr. Khwaja then joined the neurology faculty of Harvard Medical School and the neurology staff of Boston Children's Hospital, where he was director of neurogenetics. Passionate about the need to bring forward new treatment options for rare and neurodevelopmental disorders, Dr. Khwaja is on the Board and Scientific Advisory Boards of several biotechnology companies as well as patient research foundations. He is an honorary consultant pediatric neurologist to the Royal London Hospital and is a member of the Royal College of Physicians and the Royal College of Paediatrics and Child Health.



Jennifer Panagoulas, RAC
Foundation for Angelman Syndrome Therapeutics and
Angelman Syndrome Biomarker and Outcome Measure
Consortium

Jennifer Panagoulas, RAC is a Regulatory Advisor to the Foundation for Angelman Syndrome Therapeutics (FAST) and the Co-Director of the Angelman Syndrome Biomarkers and Outcome Measures Consortium (ABOM). Ms. Panagoulas has worked in drug development for over 20 years, primarily focused on advancing global development programs for the treatment of rare neurological diseases. She spent 16 years in Regulatory Affairs at Genzyme where she held various roles including Global Therapeutic Head, Regulatory Affairs Neurology. At Genzyme, she supported global registration efforts for Myozyme (alglucosidase alfa), an enzyme replacement therapy for children and adults with Pompe disease, a rare, genetic, neuromuscular disorder. Ms. Panagoulas has experience in working with global regulatory agencies including the US FDA, the European Medicines Agency, and the Pharmaceutical and Medical Device Agency in Japan. She has held key leadership roles in Regulatory Affairs supporting the development of oligonucleotide drugs at Alnylam Pharmaceuticals and Wave Life Sciences and was part of the founding team at GeneTx Biotherapeutics advancing an ASO specific for the treatment of Angelman syndrome.

**Ben Philpot, PhD****University of North Carolina at Chapel Hill**

Dr. Ben Philpot is a Kenan Distinguished Professor in the Neuroscience Center and Department of Cell Biology & Physiology at the University of North Carolina. He earned his PhD in psychobiology from Dr. Peter Brunjes at the University of Virginia and performed a postdoctoral fellowship in the laboratory of Dr. Mark Bear at Brown University and M.I.T., where he made important contributions to our understanding of experience-dependent brain

development. He is currently the Associate Director of the UNC Neuroscience Center and a member of the Carolina Institute for Developmental Disabilities, for which he helps direct a cross-disciplinary postdoctoral training grant for neurodevelopmental disorders. Dr. Philpot's current research seeks to understand the pathophysiology underlying monogenic neurodevelopmental disorders, and he uses this information to develop small molecule and gene therapies to treat these disorders. His research focuses on early-stage development of treatments for Pitt-Hopkins, Dup15q, and Angelman syndromes. Dr. Philpot has made key therapeutic discoveries, including developing an approach to unsilence the epigenetically-repressed paternal UBE3A allele as a novel treatment strategy for Angelman syndrome. Dr. Philpot has >90 peer-reviewed scientific publications. He has advised prominent biotech and pharmaceutical companies, and serves on the scientific advisory committee for the Angelman Syndrome Foundation. He has won multiple awards, including the NARSAD Young Investigator Award, a Whitehall Foundation fellowship, and the Dr. Claudia Benton Award for Scientific Research, and is currently a SFARI Investigator of the Simons Foundation.

**Mustafa Sahin, ScB, MD, PhD****Boston Children's Hospital and Harvard Medical School**

Dr. Mustafa Sahin is a developmental neurobiologist and a pediatric neurologist at Boston Children's Hospital and Harvard Medical School. He received his ScB degree from Brown University, his MD and PhD from Yale School of Medicine. He completed a pediatrics residency at Children's Hospital of Philadelphia and a child neurology residency at Boston Children's Hospital. Dr. Sahin is a Professor at Harvard Medical school and the Rosamund

Stone Zander Chair at Boston Children's Hospital. At Boston Children's, Dr. Sahin is the Director of the Translational Research Program and the Translational Neuroscience Center. Dr. Sahin has established and directs the Multidisciplinary Tuberous Sclerosis Program. He directs a national consortia to study biomarkers and comparative pathobiology of TSC and related neurodevelopmental disorders.



David Jay Segal, PhD
University of California, Davis

Dr. David Segal is a Professor at the Genome Center, Biochemistry and Molecular Medicine, Pharmacology, MIND Institute. He is also Co-Chair of the Integrative Genetics and Genomics graduate group and the Specialty Chief Editor for Frontiers in Genome Editing: Neurologic Disorders. Dr. Segal received his PhD from the University of Utah and performed a post-doc at The Scripps Research Institute. Now, at UC Davis, Dr. Segal's research focuses on gene and epigenome editing to treat Angelman syndrome and related neurologic disorders. He is a founding member of the Center for Interventional Genetics at the UC Davis MIND Institute, and is an investigator in the NIH Somatic Cell Genome Editing Consortium.



Elliott Sherr, MD, PhD
University of California, San Francisco

Dr. Elliott Sherr is a Professor in Neurology and Pediatrics at the Weill Institute of Neurosciences and the Institute of Human Genetics at UCSF. He directs the Brain Development Research Program, a group that studies the genetics and biology autism and epilepsy. Specific areas of interest include understanding the link between advance brain imaging metrics, blood-based biochemical biomarkers and autism susceptibility. His lab also studies how brain

function is altered in a “genetics first” model of ASD, as exemplified by deletion or duplication of a 600 kb interval in 16p11.2, the most common genetic cause of autism. He studies the genetics of disorders of brain development, including agenesis of the corpus callosum. In this process, he has identified several genes that are mechanistically linking these to autism. Dr. Sherr is also a member of a large epilepsy genetics consortium in which he leads a team trying to understand the genetic causes of severe childhood epilepsies, such as infantile spasms. For his research, he was the 2006 recipient of the Philip R. Dodge Young Investigator Award from the Child Neurology Society. He is a board-certified Child Neurologist and codirects the Comprehensive Center for Brain Development at UCSF. In this capacity, he cares for children with neurodevelopmental disorders, including autism, intellectual disability and epilepsy. He is a native of California and completed his undergraduate degree in Philosophy and Biology at Stanford University. He obtained his MD and PhD at Columbia University in New York and completed his clinical training in Pediatrics and Neurology at UCSF.



John E Spiro, PhD
Simons Foundation

Dr. John Spiro is the Deputy Scientific Director of the Simons Foundation Autism Research Initiative (SFARI). The Simons Foundation is a private non-profit foundation dedicated to advancing the frontiers of research in the basic sciences and mathematics. The mission of SFARI(SFARI.org), a division within the Simons Foundation with a budget of ~\$100 million/year, is to improve the understanding, diagnosis and treatment of autism spectrum disorders by funding innovative research of the greatest quality and relevance. Dr. Spiro joined the foundation in 2007 and works with the Director to oversee all aspects of the Foundation's autism research initiatives; he was interim Director from Oct 2020 to September 2021. He has also been involved in efforts to promote the use of preprints in the life sciences as well as other initiatives aimed at more open data sharing. Dr. Spiro earned his undergraduate degree from Haverford College, his PhD in neuroscience from the University of California, San Diego, and he did postdoctoral work at Duke University. Before joining SFARI, Dr. Spiro worked at Nature Publishing Group as an editor at Nature Neuroscience and then as a senior editor on the biology team of Nature, where he oversaw a group of editors responsible for editorial decisions and peer review of manuscripts across all areas of neuroscience.



Matthew State, MD, PhD
University of California, San Francisco

Matthew State MD, PhD is the Oberndorf Family Distinguished Professor of Psychiatry, Chair of the Department of Psychiatry and Behavioral Sciences, President of the Langley Porter Psychiatric Hospital and Clinics, and member of the Weill Institute for Neurosciences at the University of California San Francisco. He is a board certified child and adolescent psychiatrist and human geneticist. Over the past 15 years, his laboratory has played a key role in elaborating the contribution of rare and de novo mutations to the etiology of autism spectrum and Tourette disorders. His lab has also been a leader in using systems biological approaches to identify shared pathology among functionally diverse large effect risk genes. Dr. State has been the recipient of multiple awards, including the Ruane Prize from the Brain and Behavior Research Foundation and the Sarnat International Prize in Mental Health from the US National Academy of Medicine. He was elected to membership in the National Academy of Medicine (NAM) in 2013.



Yael Weiss, MD, PhD
Mahzi Therapeutics

Dr. Yael Weiss completed her MD, PhD at Hadassah Medical School at the Hebrew University in Jerusalem. She has over 20 years of industry experience in medical/clinical and business development roles at Genzyme, Merck and Ultragenyx. Dr. Weiss founded Mahzi Therapeutics in 2020 to bring therapies to patients with underdiagnosed ultra-rare genetic neurodevelopmental disorders. Mahzi works closely with patient foundations to support their journey towards drug development, and bring programs into Mahzi once pre-clinical proof of concept is established. Dr. Weiss is a member of the NIH driven Bespoke Gene Therapy (BCTG) consortium, ASGCT translational committee, N=1 collaborative. She is also a board member/advisor to ADNP and FOXP1 foundations.



James Wilson, MD, PhD
University of Pennsylvania

James Wilson, MD, PhD, is the Rose H. Weiss Professor and Director; Orphan Disease Center Professor of Medicine and Pediatrics; Director, Gene Therapy Program Perelman School of Medicine, University of Pennsylvania. Dr. Wilson is also the Co-Founder and Scientific Advisor, Scout Bio; Co-Founder and Chief Scientific Advisor, Passage Bio; Co-Founder G2 Bio Co-Founder; Chief Scientific Officer, and Board Member of Institute for Life-Changing Medicine (ILCM); Co-Founder and Chief Scientific Advisor, iECURE; and Chief Scientific Advisor, Center for Breakthrough Medicines. Dr. Wilson is a Professor in the Perelman School of Medicine at the University of Pennsylvania where he has led an effort to develop the field of gene therapy. His research career spanning over 40 years has focused on rare diseases and ways to treat them by gene therapy. Dr. Wilson has published over 600 papers and is named on over 200 patents worldwide. The Wilson lab identified a new type of vector based on novel isolates of adeno-associated viruses which have become best in class for gene therapy. More recently Dr. Wilson's laboratory has focused on improved vectors for gene therapy and clinical applications of genome editing and mRNA therapy.



Ashley Winslow, PhD
Odylia Therapeutics

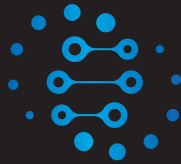
Dr. Ashley Winslow is President and Chief Scientific Officer of Odylia Therapeutics, a nonprofit biotech focused on developing gene therapies for rare diseases. Odylia develops therapeutics in collaboration with patient groups and a strategic network of CROs, academic partners, and industry, with the aim of bringing life changing therapeutics to patients. Ashley received her

PhD in Medical Genetics from the University of Cambridge and completed her postdoctoral work at Massachusetts General Hospital and Harvard Medical School. Before joining Odylia, Dr. Winslow worked in the Precision Medicine and Human Genetics and Computational Biomedicine group at Pfizer and the Orphan Disease Center at the University of Pennsylvania. She has experience overseeing drug development from target discovery to early phase clinical programs and brings to Odylia more than 15 years of drug development experience in academic, industry, and non-profit sectors. Dr. Winslow serves on the International Angelman Syndrome Research Council (INSYNC-AS) and the CHAMP1 Scientific Advisory Board.

**Mark Zylka, PhD****The University of North Carolina at Chapel Hill**

Dr. Mark Zylka is the Director of the UNC Neuroscience Center W.R. Kenan, Jr. Distinguished Professor, Department of Cell Biology & Physiology at the University of North Carolina at Chapel Hill. Dr. Zylka received a BS in Biochemistry from Virginia Tech and a PhD in Neurobiology from Harvard University. Dr. Zylka's lab is focused on developing novel treatments for chronic pain and autism. Specific projects in the lab include use of single-cell sequencing to study the normal and diseased brain, using machine learning approaches to assess pain in animal models, developing a novel CRISPR/Cas9-based treatment for Angelman syndrome, and studying genetic and environmental risks for autism.

SPEAKERS





Ganna Balagura, MD, PhD
Vrije Universiteit Amsterdam

Dr. Ganna Balagura is a certified MD in Italy and she obtained a PhD in Child Neurology at the Pediatric Neurology and Muscular Disease Unit at G. Gaslini Institute. Her research is focused on Epilepsies with genetic etiology, from bench to bedside. She is deeply involved in the laboratory projects to study epilepsy genes and targeted therapies in cellular models derived from patients, as well as in clinical studies aimed at deep phenotyping of rare disorders in order to better define diagnosis, prognosis and therapy testing. To this aim, Dr. Balagura is also actively collaborating with ILAE Epilepsiome Task Force and SNOMED CT. She is currently working as a Post-doc at Functional Genomics Department of Vrije Universiteit Amsterdam, under the supervision of Prof. Ruud Toonen and Prof. Matthijs Verhage. The projects involve therapy testing rescue the STXBP1 haploinsufficiency and building a European STXBP1 working group to improve our understanding of STXBP1 related disorders and possible targeted treatments.



Matthew Bellgard, PhD
Queensland University of Technology

Professor Bellgard is the inaugural eResearch Director at Queensland University of Technology, leading the eResearch Office to work closely with researchers and external end users to understand the research question, the breadth of technical and socio-technical challenges in order to devise innovative strategies to ultimately ensure technological solutions are fit-for-purpose and best-of-breed. With a PhD in Computer Science from the University of Western Australia (1994) specializing in Artificial Intelligence, Professor Bellgard has personally attracted over \$44m in research funding, co-inventor of 5 full/20 provisional patents, led the design and commissioning of a world's top 100 supercomputer, co-authored over 139 peer reviewed articles in areas including human/animal/plant genomics, bioinformatics, health informatics, AI, biosecurity, eResearch, remote sensing, music and radio astronomy. He has led the design and development of digital health solutions for government, industry and academia, and higher education Board member of the APEC Life Science Innovation Forum Executive Board, Chair of the APEC LSIF Rare Disease Network. Professor Bellgard is also a Board member of QCIF Ltd.

**Lynne Bird, MD****Rady Children's Hospital; University of California, San Diego**

Dr. Lynne Bird is a dysmorphologist and clinical geneticist, and Professor of Clinical Pediatrics at University of California San Diego, based at Rady Children's Hospital San Diego. She has been in practice >25 years and has been involved with Angelman syndrome research for 20 years. She has personally evaluated more than 150 individuals with Angelman syndrome. Dr. Bird is a principal investigator on several clinical trials aimed at finding treatments for the unmet needs in Angelman syndrome.



Michael Boland, PhD
Columbia University Medical Center

Dr. Michael Boland is an Assistant Professor in the Department of Neurology and the Institute for Genomic Medicine at Columbia University Medical Center. His group takes an integrated developmental and functional approach to study genes associated with pediatric epileptic encephalopathy and autism spectrum disorder using genetically engineered human stem cell and genetic mouse models. Their multi-modal approach to disease modeling combines morphological and microcircuit studies, neuronal network and animal behavior, and transcriptomic and gene network analyses to identify and understand mutation-specific phenotypes underlying seizures and cognitive impairment. They are particularly interested in drug repurposing and gene targeted approaches for preclinical therapeutic development. His group has developed patient-specific hiPSC models of Fragile X Syndrome, and organoid models of cortical malformations of development attributed to MAP1B and FLNA mutations, as well as studied neurodevelopmental disorders that result from mutations in GRIN2A, KCNT1, GNB1, and HNRNPU, and disorders of glycosylation/deglycosylation (PMM2, DPAGT1, NGLY1, SLC35A2, and ALG13). Recently his research has focused on STXBP1 encephalopathy after his son infant son was diagnosed with a pathological variant in STXPB1. Dr. Boland obtained his PhD in biochemistry and molecular biology at the University of Nebraska Medical Center where he discovered a new link between epigenetics and DNA repair in embryonic stem cells. He did his postdoctoral work at the Scripps Research Institute, where he studied the developmental potential and genomic structural variation of mouse pluripotent stem cells via mouse cloning and whole genome sequencing. He also studied the integration of gene expression and epigenetic abnormalities during early neurodevelopment in a human pluripotent stem cell model of fragile X syndrome. Dr. Boland joined the faculty of Columbia University in 2015.



Randall Carpenter, MD
Rett Syndrome Research Trust

In addition to Dr. Carpenter's role at the Rett Syndrome Research Trust, he is currently on the advisory boards of EU-AIMS and the Translational Neuroscience Center of Boston Children's Hospital, a research affiliate in the Department of Brain and Cognitive Sciences at MIT, and co-founder of Allos Pharma. Prior to RSRT, Dr. Carpenter co-founded Seaside Therapeutics to develop therapeutics capable of correcting molecular perturbations that increase vulnerability to intellectual disability and autism. While in industry, he led translational medicine teams responsible for eight successful IND submissions and dozens of FDA-compliant clinical trials. Prior to joining industry, Dr. Carpenter has held academic faculty appointments at Virginia Mason Medical Center, the University of Washington, and Wake Forest University.



Wendy Chung, MD, PhD
Columbia University; Simons Foundation

Wendy Chung, MD, PhD. is a clinical and molecular geneticist and the Kennedy Family Professor of Pediatrics in Medicine and Director of Clinical Genetics at Columbia University and Director of Clinical Research at SFARI/Simons Foundation. Dr. Chung directs NIH funded research programs in human genetics of many conditions including autism and neurodevelopmental disorders.

She is a national leader in the ethical, legal, and social implications of genomics. She leads the Precision Medicine Resource in the Irving Institute and the National Organization of Rare Disorders Center of Excellence at Columbia University. She received the NY Academy of Medicine Medal for Distinguished Contributions in Biomedical Science, the Rare Impact Award from the National Organization of Rare Disorders, and is a member of the National Academy of Medicine and the American Academy of Physicians. Dr. Chung received her BA in biochemistry from Cornell University, her MD from Cornell University Medical College, and her PhD from The Rockefeller University in genetics.



Gari Clifford, DPhil
Emory University

Dr. Gari Clifford is a tenured Professor of Biomedical Informatics and Biomedical Engineering at Emory University and the Georgia Institute of Technology, and the Chair of the Department of Biomedical Informatics (BMI) at Emory. His research applies signal processing and machine learning to medicine to classify, track and predict health and illness. His focus research areas include critical care, digital psychiatry, global health, mHealth, neuroinformatics and perinatal health. After training in Theoretical Physics, he transitioned to AI and Engineering for his doctorate at the University of Oxford in the 1990's. He subsequently joined MIT as a postdoctoral fellow, then Principal Research Scientist where he managed the creation of the MIMIC II database, the largest open-access critical care database in the world. He later returned to Oxford as an Associate Professor of Biomedical Engineering, where he helped found its Sleep & Circadian Neuroscience Institute and served as Director of the Centre for Doctoral Training in Healthcare Innovation at the Oxford Institute of Biomedical Engineering. Dr Clifford is a strong supporter of commercial translation, working closely with industry as an advisor to multiple companies, co-founding and serving as CTO of an MIT spin-out 'MindChild Medical' since 2009, and co-founding and serving as CSO for Lifebell AI since 2020. As Chair of BMI, Dr Clifford has established the department as a leading center for critical care and mHealth informatics, and as a champion for open-access data and open-source software in medicine, particularly through his leadership of the PhysioNet/ CinC Challenges and contributions to the PhysioNet Resource. He is committed to developing equitable and sustainable solutions to healthcare problems in resource poor locations, with much of his work focused in Guatemala.



Monica Coenraads, MBA
Rett Syndrome Research Trust

Monica Coenraads's involvement with Rett syndrome began the day her then-two-year-old daughter was diagnosed with the disorder. A year later, in 1999, she co-founded the Rett Syndrome Research Foundation (RSRF) and held the position of scientific director during the eight years of the Foundation's drive to stimulate scientific interest and research in Rett syndrome, culminating with the groundbreaking work in 2007 which demonstrated the first global reversal of symptoms in preclinical models of the disorder. Monica Coenraads launched the Rett Syndrome Research Trust in late 2008 to pursue the next steps from that milestone. As chief executive officer she oversees all aspects of the organization, including day-to-day operations, strategic direction, fundraising, and communications. Together with her colleagues and with input from advisors and the scientific community at large, she sets and executes RSRT's research agenda. In 2010 she co-founded the Rett Syndrome Research Trust UK and serves as a trustee (the organization is now called Reverse Rett). In 2013 she joined the Advisory Council for The Research Acceleration and Innovation Network (TRAIN) of FasterCures. Monica Coenraads is also a member of the Patients Count Leadership Council. In 2016 she was invited to join the UNC Autism Research Center Advisory Board. She is a founding trustee of the American Brain Coalition. She was awarded Redbook magazine's Mother & Shaker Award, alongside Katie Couric and Matilda Raffa Cuomo. In 2006, the Howard Hughes Medical Institute highlighted her efforts in the November issue of the HHMI Bulletin.

**Meagan Cross**

**Foundation for Angelman Syndrome Therapeutics,
Foundation for Angelman Syndrome Therapeutics
Australia, Global Angelman Syndrome Registry**

Shortly after Meagan Cross's youngest daughter's diagnosis with the rare neurogenetic condition Angelman syndrome, she knew that she wanted to be involved in helping to facilitate research into the syndrome and advocate for rare diseases in Australia. In 2009, Ms.

Cross joined the Foundation for Angelman Syndrome Therapeutics,

is a Cofounder of the Foundation for Angelman Syndrome Therapeutics Australia and worked on the development of the Foundation's major project, the Global Angelman Syndrome Registry, designed to address the unmet need for the large amounts of data required to understand the syndrome across geographical and socioeconomic boundaries. In addition, Ms. Cross runs a small charity with some other local "rare" mums, in her home town of Cairns, Rare Friends FNQ, that hosts an annual fun run and walk to raise awareness for Rare Disease Day. She is a member of Rare Voices Australia. A sea change to Cairns resulted in a new career pathway in Geographic Information Science. Ms. Cross runs a small geographic information science consultancy and she has experience in data development, database and website design, data management, programming, analysis, and presentation. She also manages a highly customized stainless steel fabrication business in Far North Queensland with her husband, Rohan.



Victor Faundez, MD, PhD
Emory University School of Medicine

Dr. Victor Faundez received his MD and PhD from Pontificia Universidad Catolica: Santiago de Chile, CL. He is a Professor at the Emory University School of Medicine. The Faundez Laboratory (<https://www.faundezlab.org/>) is a fundamental cell biology laboratory focused on unraveling synaptic mechanisms using molecular systems biology, biochemical, functional, and genetic approaches. They focus on the biology of rare human

neurodevelopmental Mendelian genetic disorders. They reasoned these disorders are a fertile ground to uncover novel aspects of neuronal cell biology required for synapse function. They study the 22q11.2 microdeletion syndrome, Rett syndrome, and CDKL5 disorder in order to capture diverse mechanisms of synapse dysfunction in rare disorders. All these diseases share partially overlapping neurodevelopmental and synaptic phenotypes. Thus, the Faundez Lab hypothesize that these rare disorders may share fundamental molecular disease mechanisms required for synapse function. To study these diseases and their molecular commonalities, they have developed systems biology approaches of tissues, cells, and biofluids from preclinical mouse models to identify mechanisms of disease and biomarkers. Their studies are unveiling disease pathogenic mechanisms and discovering molecules with biomarker potential for these neurodevelopmental disorders.



John Foxe, MS, PhD
University of Rochester

Dr. John Foxe is the Kilian J. and Caroline F. Schmitt Chair in Neuroscience, Director of The Del Monte Institute for Neuroscience, and serves as Chair of the Department of Neuroscience at The University of Rochester. His research investigates the neurobiological bases of neurodevelopmental and neuropsychiatric conditions such as Autism and Schizophrenia. He uses electrophysiological and neuroimaging techniques to understand

how inputs from the various sensory systems are combined in the brain, and what happens when these multisensory integration abilities are impacted by disease. Dr. Foxe has authored more than 300 research and clinical papers, book chapters, commentaries, and proceedings and serves as editor-in-chief of The European Journal of Neuroscience. Before joining the University of Rochester in 2015, he was director of research for the Children's Evaluation and Rehabilitation Center at the Albert Einstein College of Medicine. Originally from Dublin Ireland, Dr. Foxe read English and History at University College Dublin (1987) before completing his BS degree at Iona College, New Rochelle (1989). He obtained his MS and PhD in Neuroscience from the Albert Einstein College of Medicine in New York City (1999).



James Goss, PhD
STXBP1 Foundation

Dr. James Goss is a neuroscientist with 30 years of experience in academia, bio/pharma, and non-profits primarily focused on neurologic pre-clinical drug development. He has carried out basic research on aging, neurodegeneration, traumatic brain injury, and genetics. He collaborated in the development of a gene therapy platform based on herpes simplex virus and developed gene therapies for several neurological disorders including

Parkinson's disease, chronic pain, and peripheral neuropathy, including the first clinical trial for an HSV-based therapeutic. He has spent the last several years working with non-profit organizations, first in developing anti-HIV drug/device combinations then managing the gene therapy program for neurofibromatosis type 1. Dr. Goss is currently the Scientific Director for the STXBP1 Foundation where he is using his knowledge and experience in neuroscience and drug development to meet the challenges ahead in finding a cure.



Zachary Grinspan, MD, MS
Weill Cornell Medicine

Dr. Zachary Grinspan is an Associate Professor in Pediatrics and in Population Health Sciences, Interim Chief of Child Neurology, and Director of Pediatric Epilepsy at Weill Cornell Medicine in New York City. He has funded and led several multi-institutional research initiatives, including the CDC-funded Rare Epilepsies in New York City project. He is the primary investigator for the Pediatric Epilepsy Learning Healthcare System, a consortium of more than 20 pediatric epilepsy centers that aims to reduce seizures and their consequences for children with epilepsy through cycles of health data collection and analysis, dissemination of new evidence, and practice change. Dr. Grinspan currently leads a pilot clinical trial of glycerol phenylbutyrate for STXBP1 and SLC6A1 (NCT04937062).



Rachael Hawtin, PhD
Ultragenyx Pharmaceutical Inc.

Dr. Rachael Hawtin has been involved in drug development with a focus on translational sciences for over 25 years, predominantly in oncology, immuno-oncology and autoimmunity, and more recently rare diseases including neurologic indications. Throughout her career she has worked to better understand the pathobiology of disease and mechanism-of-action of therapeutics, to enable rationale-based target selection and combination approaches, and to better pair patients with treatments. Her leadership roles have encompassed research target identification and biomarker development through to leadership of phase 2 and 3 clinical studies. Dr. Hawtin is currently the Executive Director of Biomarker Strategy at Ultragenyx, a company focused on rare and ultra-rare genetic diseases. Ultragenyx is exploring multiple avenues to apply biomarkers to accelerate drug development in these populations and deliver therapeutics to patients in need. Ultragenyx is working in partnership with GeneTx Biotherapeutics to develop GTX-102 for treatment of Angelman syndrome. She has worked at SUGEN, Chiron / Novartis, Sunesis pharmaceutical, Nodality, Gilead Sciences, and currently at Ultragenyx.



Ingo Helbig, MD
Children's Hospital of Philadelphia

Dr. Ingo Helbig is a child neurologist and epilepsy genetics researcher at the Children's Hospital of Philadelphia. He received his MD from the University of Heidelberg, Germany, and completed his pediatric and child neurology residency in Melbourne (Australia), Kiel (Germany), and Philadelphia (USA). He is currently an Assistant Professor of Neurology and Pediatrics at the Perelman School of Medicine at the University of Pennsylvania and The Children's

Hospital of Philadelphia. His group has contributed to the major gene discoveries in human epilepsies over the last decade and is currently expanding its focus to apply data-driven approaches to understand epilepsy phenotypes. Dr. Helbig co-leads the Epilepsy Gene Curation Panel of the ClinGen consortium and is the Director of Genomic Science and Fellowship Director of the Epilepsy NeuroGenetics Initiative at Children's Hospital of Philadelphia, one of the world's largest epilepsy genetics programs with more than 1,000 new epilepsy genetics evaluations annually. Dr. Helbig is also well known for his neurogenetics blog "Beyond the Ion Channel" (www.epilepsygenetics.net).



Honey Heussler, MD
Children's Health Queensland Hospital

Dr. Honey Heussler is the Medical Director of the Child Development Program at the Children's Health Queensland Hospital and Health Service in Brisbane. She is a Developmental and Behavioral Pediatrician with a dual qualification in Sleep Medicine. Dr. Heussler's clinical work involves children with a variety of Developmental and Behavioral problems as well as a number of clinics that specialize in sleep disorders for this population. She also runs a specialized clinic for some genetic disorders including Angelman syndrome. Dr. Heussler is a member of the Scientific Advisory Board for the Foundation for Angelman Syndrome Therapeutics and has been heavily involved in the Angelman syndrome community for over a decade.

**Joerg Hipp, PhD**
Roche

Dr. Joerg Hipp is a biomarker and experimental medicine leader at Roche. He has a background in Physics and Neuroscience. Before joining Roche, his main research focus was to understand the role of neuronal oscillations in normal and pathological brain function using different techniques including EEG, MEG and MRI. At Roche, Dr. Hipp is responsible for the biomarker strategy and implementation in several neurodevelopmental disorder programs and heads a group that bundles clinical electrophysiology and sleep expertise to support the neuroscience portfolio.



Dina Katabi, MS, PhD
Massachusetts Institute of Technology

Dr. Dina Katabi is the Thuan and Nicole Pham Professor of Electrical Engineering and Computer Science at MIT. She is also the director of the MIT's Center for Wireless Networks and Mobile Computing, a member of the National Academy of Engineering, and a recipient of the MacArthur Genius Award. Professor Katabi received her PhD and MS from MIT in 2003 and 1999, and her Bachelor of Science from Damascus University in 1995. Dr. Katabi's research focuses on innovations in digital health, applied machine learning and wireless sensors and networks. Her research has been recognized with ACM Prize in Computing, the ACM Grace Murray Hopper Award, two SIGCOMM Test-of-Time Awards, the Faculty Research Innovation Fellowship, a Sloan Fellowship, the NBX Career Development chair, and the NSF CAREER award. Her students received the ACM Best Doctoral Dissertation Award in Computer Science and Engineering twice. Further, her work was recognized by the IEEE William R. Bennett prize, three ACM SIGCOMM Best Paper awards, an NSDI Best Paper award and a TR10 award. Several start-ups have been spun out of Dr. Katabi's lab such as PiCharging and Emerald.



Alana Newhouse
Foundation for Angelman Syndrome Therapeutics
Tablet Magazine

Alana Newhouse is the editor-in-chief of Tablet Magazine, which she founded in 2009. Originally established as an outlet devoted to covering Jewish life, Tablet is now into one of the most influential platforms for news and ideas on the web, an address for reporting and arguments regularly cited by the New York Times, the Washington Post, Associated Press, The New Yorker, and others—read by readers of all backgrounds, from around the world. But her most important job is that she is the mother of Elijah, 7, who has a mutation in the UBE3A gene resulting in Angelman syndrome.

**Laurent Servais, MD, PhD****MDUK Oxford Neuromuscular Centre; University of Liège**

Dr. Laurent Servais is a Professor of Paediatric Neuromuscular Diseases at the MDUK Oxford Neuromuscular Centre and Invited Professor of Child Neurology at Liège University. After graduating from Louvain Medical School, Brussels, Belgium in 1999, Dr. Servais completed a PhD in Neuroscience about cerebellar electrophysiology in alert mice, including in an Angelman mouse model from Free University of Brussels, Belgium. It was followed

by residencies in child neurology at the Free University of Brussels and Robert Debré Hospital, Paris. In 2008, he took a position in neuromuscular disease and clinical research at the Institute of Myology in Paris, where his interest and expertise in neuromuscular diseases flourished. Dr. Servais was subsequently appointed Head of Clinical Trials and Database Services. Most recently, he served as Head of the Institute of Myology's I-Motion (Institute of Muscle-Oriented Translational Innovation), and Head of the Neuromuscular Centre in Liège, Belgium. He joined the MDUK Oxford Neuromuscular Centre and the University of Oxford in September 2019. Dr. Servais' main interest is about newborn screening, innovative clinical trials design and new outcome measures based on wearable technology. This is actually how he came back in the Angelman field, being deeply involved in the intellectual process of the development of new digital outcomes in Angelman syndrome.



Charlene Son Rigby, MBA
RARE-X and STXBP1 Foundation

Charlene Son Rigby is the Chief Executive Officer of RARE-X. She has spent her career building organizations at the intersection of data, technology, and life sciences. Charlene was previously Chief Business Officer at Fabric Genomics and held executive roles at enterprise software and genomics companies, including Oracle and DoubletWist. She started her career in neuroscience research at Roche. When Charlene's daughter was diagnosed with a rare genetic disease, she co-founded the STXBP1 Foundation. She is committed to finding a cure for her daughter's disorder. Charlene's unplanned connection between her personal life and profession has helped push forward the search for a cure for her daughter and kids like her, and given her work deeper meaning. She holds a BA in Human Biology from Stanford University and an MBA from the Haas School of Business at U.C. Berkeley.



Dudley Tabakin, MSc
VivoSense

Dudley Tabakin, MSc, is the CEO and co-founder of VivoSense and a fervent believer in “good data” over “big data” in the development of digital clinical measures from wearable sensor technology. He has a background in sports and gait biomechanics and more than 10 years experience in wearable sensor data analytics.



Matthijs Verhage, PhD

Vrije Universiteit & Amsterdam University Medical Center

Dr. Matthijs Verhage is a synapse biologist based in Amsterdam. He pioneered the utilization of stem cell technology for synapse research and for disease modelling in neurodevelopmental disorders. His lab focusses on SNAREopathies, severe neurodevelopmental disorders caused by mutations in the eight genes that together form the core machinery for the secretion of chemical signals in the synapse. His team develops new therapies

for SNAREopathies together with private companies and works together with clinicians and patient families worldwide to promote trial readiness. Matthijs Verhage is professor head of the Functional Genomics Department at the Center for Neurogenomics and Cognitive Research (CNCR), Vrije Universiteit & Amsterdam University Medical Center, Amsterdam, The Netherlands. He is also affiliated with Karolinska Institute, Stockholm, Sweden (SUN) and the Broad Institute, MIT Cambridge MA, USA (SYNGO). He obtained his PhD at the University of Amsterdam in 1990 (cum laude) and received post-doctoral training at the labs of prof. David G. Nicholls (Dundee, UK) and the Nobel laureate prof. Thomas C. Südhof (Howard Hughes Medical Institute, Dallas, USA). Matthijs Verhage was partner of the EU FP6 consortium EU-Synapse, FP7 consortia EuroSpin and SynSys, co-founder and vice chair of H2020 consortium COSYN, and co-founder/chair of the SYNGO consortium (together with Guus Smit), funded by The Broad Institute/MIT, the BRAINMODEL consortium awarded by the Dutch national grant authority ZonMW in 2021 and founder/chair of the ESCO consortium to promote trial readiness for SNAREopathies in Europe. In 2013 he received the ERC Advanced Grant of the European Research Council. Matthijs Verhage is chair/member of scientific advisory/review boards of the IPNP (Descartes), Paris, France; the STXBP1 foundation (patient organization, NC, USA); the IBS Center for Synaptic Brain Dysfunctions, Deajeon, Korea, the Centro de Biología Molecular Severo Ochoa, Madrid Spain and the CIBB, Coimbra, Portugal. Matthijs Verhage is inventor on patent applications related to gene therapy and he is founder/director of Alea Biotech, Sylics, and Neurospecter and consultant/scientific advisor for Life Science companies in The Netherlands, USA, Spain and Japan.



Jana von Hehn, PhD
Rett Syndrome Research Trust

Dr. Jana von Hehn is a molecular biologist and geneticist who devoted her academic career to the study of pediatric genetic disorders including fragile X syndrome, galactosemia, and ataxia-telangiectasia at Emory University and Yale. Dr. von Hehn has worked within the biopharmaceutical industry in clinical development planning and clinical trial operations in the areas of pediatric and adult seizure disorders, fragile X syndrome, autism spectrum disorder, as well as solid and liquid tumors, autoimmune, and respiratory disorders. With 15 years of experience in drug development under 13 Investigational New Drug applications, she ran pivotal studies that contributed to a drug approval by the FDA in 2019, and is an inventor on a patent for a small molecule currently in development. Prior to pursuing her PhD, Jana earned her BS in molecular biology and microbiology at the University of Central Florida. As an undergraduate, she earned a scholarship for the pursuit of higher education from the Miss America Organization when she won the title of Miss University of Central Florida 2000 with her platform for advancing genetic research to treat disease. She is the daughter of a major league baseball pitcher and was born and raised in sunny south Florida with her three sisters. When not working to cure Rett syndrome, Jana enjoys cooking, sports, gardening, comedies, and spending time with her friends and family. She currently resides in greater Boston.



Paul Wang, MD
Simons Foundation

Dr. Paul Wang is a developmental-behavioral pediatrician who has worked in academia, industry, and non-profit sectors, always with a focus on neurodevelopmental disorders. While serving on the faculty at Children's Hospital of Philadelphia, he helped to care for hundreds of children and families affected by genetic diagnoses, autism, and intellectual disabilities. His research there explored language and memory development in these conditions. Dr. Wang subsequently worked at Pfizer and then at Seaside Therapeutics, where he led drug development efforts for autism and Fragile X Syndrome. He subsequently served as Vice President for Clinical Affairs at Autism Speaks, and now is Deputy Director for Clinical Research at the Simons Foundation (SFARI).

**Anne Wheeler, PhD****RTI International; University of North Carolina at Chapel Hill**

Dr. Anne Wheeler is a senior research analyst at RTI International, where she conducts multiple research projects focused on the development of tools and strategies to improve outcomes for individuals with intellectual/developmental disabilities, especially those with rare neurogenetic conditions. She has been involved in research on focused on individuals with neurogenetic conditions and their families for over 20 years. She is the director of the

LADDER database, is working to identify and improve outcome measures used to determine change in clinical trials for rare conditions, and leads efforts to reduce the age of diagnosis and provide targeted early intervention and support services for children diagnosed in infancy. Dr. Wheeler is also an Adjunct Associate Professor of Psychiatry and School Psychology at the University of North Carolina at Chapel Hill and a practicing licensed psychologist at the Carolina Institute for Developmental Disabilities where she provides developmental and behavioral consultation for individuals with Angelman and Dup15q syndromes. She has been involved in clinical service provision and research focused on developmental and familial outcomes for children with genetic diagnoses for over 15 years.

The background is a solid dark blue. It features several light blue circular elements of varying sizes. Three prominent horizontal bars, each consisting of two light blue circles connected by a thin line, are arranged vertically. Other smaller circles are scattered throughout the composition.

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