FAST Translational Research Symposium and ABOM Scientific Meeting Program





FAST Translational Research Symposium and **ABOM Scientific Meeting** Thursday, December 1, 2022 8:30 am – 5:45 pm

Welcome to The Angelman Syndrome Biomarker and Outcome Measure Consortium Scientific Meeting and the Translational Research Symposium for the Foundation for Angelman Syndrome Therapeutics (FAST).

We are incredibly grateful that so many of you are joining us as we build a community of scientists, clinicians, drug developers, regulators, and other thought leaders, to accelerate drug development for Angelman syndrome.

Our hope is that these next few days will help to advance the drug development pipeline for Angelman syndrome and ensure we are doing all we can to support the needs of translational research, clinical trial readiness and future therapeutic approvals.

The progress in this field has been mind-blowing, but we also realize that these are still early days. By bringing diverse talent together, like each of you here, we hope to encourage and accelerate the highest quality work to bring the most meaningful change to the lives of all individuals living with Angelman syndrome.

Thank you for being a part of this incredible collaborative effort!

Allyson Berent, DVM, DACVIM

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

Jennifer Panagoulias, RAC

Head of Regulatory and Compliance, Foundation for Angelman Syndrome Therapeutics Co-Director, Angelman Syndrome Biomarker and Outcome Measure Consortium (ABOM) jennifer.panagoulias@cureangelman.org

David J Segal, PhD

Co-Chair, FAST Translational Research Symposium Professor and Co-Chair of the Integrative Genetics and Genomics graduate group Genome Center, Biochemistry and Molecular Medicine, Pharmacology, and MIND Institute University of California, Davis djsegal@ucdavis.edu



Program Schedule

Time	Speaker/Organization	
8:30 am	Allyson Berent, DVM, DACVIM FAST	
	Jennifer Panagoulias, RAC FAST, Angelman Syndrome Biomarker and Outcome Measure Consortium	

Natural History and Real World Data

8:35 am	Dora Markati, MD, MPhil University of Oxford
8:50 am	Bridgette Kelleher, PhD Purdue University
9:05 am	Jessica Duis, MD, MSc Children's Hospital Colorado
9:20 am	Laurent Servais, MD, PhD University of Oxford
9:35 am	All Natural History and Real World Data Presenters

Clinical Outcomes and Meaning to Patients

9:50 am	Sarah Nelson Potter, PhD, CCC-SLF RTI International
10:05 am	Angela You Gwaltney, PhD RTI International
10:20 am	Anjali Sadhwani, PhD Boston Children's Hospital

Topic

Welcome

University of Oxford Angelman Syndrome Natural History Study

Characterizing the Natural History of Angelman Syndrome in Early Childhood: Initial Results from Phase I of the Purdue Early Phenotype Study (2016-2021)

The Functional Independence Measure (WeeFIM) for Individuals with Angelman Syndrome

Real-Life Assessment of Gait and Walking Behavior of Angelman Patients

Panel Discussion

Quality of Life for Families Living with Angelman Syndrome

Adaptive Functioning in Angelman Syndrome and its Relationship to Developmental Functioning

Determining Meaningful Change Thresholds for Critical COAs in Angelman Syndrome

10:40 am	Christina K. Zigler, PhD, MSEd Duke University School of Medicine	ORCA: Longitudinal Data with Associated Correlations with the BSID, VABS and Communication Matrix from the FDA Funded NHS in Angelman Syndrome		1:55 pm	Adriana Gomes, MD UCSD/Rady Children's Hospital-San Diego Kelly David FAST
11:00 am	Michelle Campbell, PhD U.S. Food and Drug Administration	How Data Inform Drug Development: A Regulatory Perspective	-		Amanda Moore Angelman Syndrome Foundation
11:25 am	All Clinical Outcomes and Meaning to Patients Presenters	Panel Discussion		2:00 pm	Rob Carson, MD, PhD Vanderbilt University Medical Center
11:40 am	Break		-	2:10 pm	All Clinical Data Presenters

Biomarkers

11:55 am	Mike Sidorov, PhD Children's National Hospital	Evaluating Peak Alpha Frequency as an EEG Biomarker for Angelman Syndrome	2:25 pm
	Abigail Dickinson, PhD		
	UCLA		
12:05 pm	Dan Foti, PhD	Telehealth EEG Assessment in Angelman Syndrome:	
	Purdue University	Preliminary Analyses of Signal Quality and Detection of Increased Delta Power	
12:15 pm	Robert Komorowski, PhD	Detecting UBE3A in the Cerebral Spinal Fluid of Patients	0.20 pm
	Ionis Pharmaceuticals	With Angelman Syndrome	2:50 pm
12:25 pm	Alexandra Key, PhD	Early Sensory vs. Later Cognitive Neural Markers of	
	Vanderbilt University Medical Center	Communicative Functioning in Angelman Syndrome	2:45 pm
12:35 pm	All Biomarkers Presenters	Panel Discussion	
12:50 pm	Lunch		3:00 pm

Translational Research Symposium

2:30 pm	Henriette O'Geen, PhD University of California, Davis Genome Center
2:45 pm	Maria Clarke, BS University of California San Francisco
3:00 pm	Ulrika Beitnere, PhD University of California, Davis Genome Center
3:15 pm	All Upstream Targeting Presenters
3:25 pm	Break

David J. Segal, PhD

University of California, Davis

Clinical Data

1:35 pm Erick Sell, MD Children's Hospital of Eastern Ontario Safety and Tolerability of GTX-102 in Pediatric Patients with Angelman Syndrome from a Phase 1/2 Open-Label, Multiple-Dose, Dose-Escalation Clinical Trial

Downstream Targeting-Preclinical Data

3:40 pm

Joerg Hipp, PhD Roche

Community-Sourced Reporting of Mortalities in Angelman Syndrome (1979-2021) Compared to Prospective Natural History And Registry Data

Crowd-Sourcing Research into Nonepileptic Myoclonus in Angelman Syndrome

Panel Discussion

Translational Research Symposium Introduction

Upstream Targeting-Preclinical Data

r	Transcriptional Reprogramming Restores UBE3A Brain-Wide and Rescues Behavioral Phenotypes in an Angelman Syndrome Mouse Model
	A Prenatal Antisense Oligonucleotide Therapy for Angelman Syndrome in a Mouse Model
r	CRISPR RNA Targeting Tools for Angelman Syndrome
	Panel Discussion

Locally Referenced Beta Power is Decreased in Deletion Angelman Syndrome and May Provide a Biomarker for Reduced GABA-A Receptor Function in this Disorder

3:50 pm	Maria-Clemencia Hernandez, PhD Hoffman-La Roche Ltd	Alogabat, a GABAA-A5 Positive Allosteric Modulator, as a Novel Therapeutic Approach to Restore Deficient	5:30 pm	Allyson Berent, DVM, DACVIM FAST	
		GABAergic Signaling in Deletion Angelman Syndrome		Jennifer Panagoulias, RAC FAST, Angelman Syndrome Biomarker and	
4:00 pm	Wayne Chadwick, PhD	AI-Powered Drug Discovery Identifies a Potential New		Outcome Measure Consortium	
	HEALX	Treatment for Angelman Syndrome		David J. Segal, PhD	
				University of California, Davis	
4:10 pm	John Marshall, PhD	Syn3 Peptidomimetics Improve Deficits in Learning in a			
	Brown University	Mouse Model of Angelman Syndrome	5:40 pm	Reception	
4:20 pm	Timothy Fenton University of California, Davis	Establishing Structural and Functional Outcome Measures in Vitro for Testing Therapeutics for Angelman Syndrome			
4:30 pm	Scott Dindot, PhD Texas A&M College of Medicine	Pig Model of Angelman Syndrome: Characterization and Future Directions		You are invited to view these addition scrolling in the Grand Ballroom Foy	
4:40 pm	Z. Begum Yagci North Carolina State University	Advancing Human Stem Cell-Derived Platforms for Angelman Syndrome Research		Deirdre Neenan-Smith, MLS Radius Health, Inc.	
4:45 pm	All Downstream Targeting Presenters	Panel Discussion		Tomas Salter-Cid	
				University of California, Davis	

Oral Poster Lightning Round

4:55 pm	Dr. M. Rance MBBS BSc(Hons), MRCS(Eng) PTC Therapeutics	Gene Therapy for Angelman Syndrome: A Preclinical Update of PTC-AS
5:00 pm	Luis Martinez, PhD Baylor College of Medicine/ Texas Children's Hospital	Long Term Phenotypic Alterations Following Early Life Febrile Seizures and Altered Neuroimmune Response in a Rat Model of Angelman Syndrome
5:10 pm	Anne C. Wheeler, PhD RTI International	Linking Angelman and Dup15q Databases for Expanded Research (LADDER): Updates and Next Steps
5:15 pm	Mindy Leffler, MEd Casimir/Emmes	The Development of Angelman Syndrome Video Assessment (ASVA) Scorecards to Quantify Clinically Meaningful Changes in Function in Individuals with Angelman Syndrome
5:20 pm	Jake Donaghue, MD, PhD Beacon Biosignals	Machine Learning and At-Home EEG Accelerate Precision Medicines for Developmental Epileptic Encephalopathies
5:25 pm	Questions on Oral Posters	

Katerina (Kate) S. Kucera RTI International

Zachary Rosinger, PhD University of California, Davis

Ulrika Bietnere, PhD University of California, Davis Genome Center

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	An Online Survey of Caregivers for Patients with Angelman Syndrome
	Screening of Cas13d and Novel Guides for Angelman Syndrome
	Behavior and Neurophysiological Phenotypes Delineate UBE3A Overexpression from Angelman Syndrome Mouse Models
	Evaluation of a Candidate High-Throughput Methylation- Based Assay for Newborn Screening for Angelman, Prader-Willi, and Dup15q Syndromes
er	Unique Features of the Gut Microbiome Characterized in Animal Models of Angelman Syndrome

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Let's meet and discuss the impact of Angelman Syndrome on motor function

Sysnav Healthcare is committed to make a meaningful contribution to the Angelman Syndrome community

Meet us!



Damien Eggenspieler Healthcare program director damien.eggenspieler@sysnav.fr

About Sysnav Healthcare

Sysnav Healthcare is a leading business unit of Sysnav, a fast-growing French independent tech company. Since the start of its activities, Sysnav Healthcare aims to unlock the potential of real-world data in the medical field by adapting extremely precise motion-capture solutions to the needs of healthcare professionals and patients. The company has already developed two wearable devices: ActiMyo® & Syde® and is the first-ever to qualify a digital endpoint (SV95c) with EMA. Sysnav Healthcare, as the pioneer and leader for movement evaluation in clinical trials, is now building the next generation of digital health technologies so that patients benefit earlier and more reliably from life-changing treatments.







Yacine Bechichi

BD manager yacine.bechichi@sysnav.fr



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We're honored to be in the company of guardians, givers, and the greatest of groundbreakers.

We are inspired and motivated by the patients, families, and care partners we serve. We strive to develop treatments, resources, and educational materials that live up to our promise and your standards and empower patients to advocate on behalf of themselves and their rare communities.

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We proudly support the Angelman syndrome community. Your tenacity motivates us to work every day to find a treatment. Together we can **boldly** transform life with Angelman syndrome.

Thank you, families, for including Ovid in this important event. We are so happy to be back together in person.

