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)
Gerald D. Fischbach Auditorium, 2nd Floor

Zoom: https://simonsfoundation.zoom.us/j/95153627462?pwd=OUswMktyaGFxa2Q4VUJEdVU4YWYyZz09

Meeting ID: 951 5362 7462
Passcode: 236606

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 Panagoulias, RAC** (FAST)
9:10 – 10:25am	**Update and Overview on Drug Development in AS**
9:10 – 10:00am	**Allyson Berent DVM** (FAST) *Landscape for Drug Development in Angelman Syndrome: Annual Update and New Initiatives*
10:00 – 10:25am	Discussion on Landscape/Patient Identification Initiative- FAST Global Search & Rescue moderated by **Jennifer Panagoulias, 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) *The Diagnostic Odyssey in Angelman Syndrome: Clinician's Perspective*
11:05 – 11:15am	**Alana Newhouse** (FAST) *Why a Diagnosis Matters: A Parent's Perspective*
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) *Incidence and Prevalence Challenges in AS: ICD-10, Claims Database, NBS, Literature Review and Available Data*

12:00 – 12:15pm	**Laurent Servais, MD, PhD** (Oxford University) *How Patient Identification Was Accelerated in Other Rare Diseases: SMA/DMD/Other*
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) *How to Leverage the Expertise of Diagnostic Vendors to Support Patient Identification for a Community and Industry*
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 Panagoulias, RAC** (FAST) *How to Collect Patient Identification Data that is Most Effective and Useful for Community and Industry*
2:25 – 2:55pm	**Anne Wheeler, PhD** (RTI) **Honey Heussler, MD** (Queensland Hospital) **Meagan Cross** (FAST-AU) **Matthew Bellgard, PhD** (Queensland University) *Leveraging Existing Databases: GASR and LADDER*
2:55 – 3:15pm	Discussion on Database Best Practices and Creating an Infrastructure Globally moderated by **Jennifer Panagoulias, 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) *Introduction to Novel Endpoints and Pharmacodynamic Biomarkers for AS*
4:15 – 4:25pm	**John Foxe, MS, PhD** (University of Rochester) *Translational Neurophysiological Markers in Neurodevelopmental Disorders*
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 Blackbarn (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) *Wearable Biosensors*
10:30 – 10:50am	**Dina Katabi, MS, PhD** (Massachusetts Institute of Technology) *Invisible Biosensors*
10:50 – 11:10am	**Gari Clifford, DPhil** (Emory University and Georgia Institute of Technology) *Combining Wearables and Nearables for Tracking Changes in Health*
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) *Molecular Mechanisms, Cellular and Animal Models*
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) *Natural History and Disease Concept Model*
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) *ESCO: an EU Consortium to Promote Trial Readiness for STXBP1 Disorders*
1:40 – 1:55pm	**Matthijs Verhage, PhD** (Vrije Universiteit & Amsterdam University Medical Center) *Connecting Cellular Phenotypes, qEEG and Clinical Symptoms*

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) *An Overview of Gene Targeted Strategies and Drug Discovery Platforms to Treat STXBP1 Haploinsufficiency*
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 Panagoulias, 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)