STXBP1 Disorders

STXBP1 RESEARCH ROUNDTABLE 2021

Join the STXBP1 Research Roundtable, where we will convene to review research progress, discuss key gaps in current knowledge, and ways to address these gaps to accelerate therapies for STXBP1.

This two-day virtual conference will bring together STXBP1 investigators and medical professionals with a strong interest in STXBP1 and related disorders.

REGISTER

Don't Miss These Speakers

Frederic Meunier, PhD
University of Queensland

Hannah Stamberger, MD, PhD
University of Antwerp

Ingo Helbig, MD
Children's Hospital of Philadelphia

Zachary Grinspan, MD, MS
Weill-Cornell Medicine

Anne Berg, PhD
Northwestern Medicine

Xavier Liogier, PhD
Ioulou Foundation

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Encoded THERAPEUTICS

18 - 19 NOVEMBER 2021

info@stxbp1disorders.org
www.stxbp1disorders.org

STXBP1 RESEARCH ROUNDTABLE 2021

Thursday, 18 November 2021

Meeting Kickoff

9:00 am ET

Welcome
Charlene Son Rigby, MBA & James Goss, PhD
STXBP1 Foundation

Meeting Goals
Ben Prosser, PhD
University of Pennsylvania

Clinical

9:20 am ET

Session Chair: Ingo Helbig, MD

Gaps in Understanding STXBP1 Phenotype
Ingo Helbig, MD
Children's Hospital of Philadelphia

Natural History of STXBP1 in Adulthood
Hannah Stamberger, MD, PhD
University of Antwerp

Dissecting the relationship between seizures and development in STXBP1-DEE
Ganna Balagura, MD, PhD
University of Genoa

Endpoint & Biomarker Development, Lessons Learned from Clinical Trials

10:25 am ET

Session Chair: Terry Jo Bichell, PhD

Rare Diseases- Rare Outcomes - Measuring “Better” for DEE Patients with Severe Impairments
Anne Berg, PhD
Northwestern Medicine

Paving the Way for Clinical Development for CDD
Xavier Liogier, PhD
loulou Foundation

Lessons Learned from 4-phenylbutyrate Clinical Trial
Zachary Grinspan, MD, MS
Weill-Cornell Medicine

Models for Therapeutic Development

11:40 am ET

Characterization of R406H Mouse Model
Rebecca Haffner-Krausz, PhD & Michael Tsoory
Weizmann Institute

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STXBP1 RESEARCH ROUNDTABLE 2021

Friday, 19 November 2021

Day 2 Kickoff

9:00 am ET

Day 1 Observations & Day 2 Goals
Ben Prosser, PhD
University of Pennsylvania

Mechanisms of Disease

9:05 am ET

Session Chair: Jacqueline Burre, PhD

Dissecting STXBP1 disease mechanism, one molecule at a time
Frederic Meunier, PhD
University of Queensland

Reduced MUNC18-1 levels, synaptic proteome changes and altered network activity in STXBP1-Syndrome patient neurons
Annemiek van Berkel and Hanna Lammertse, MSc
VU Amsterdam

Multiplicative effects of a modifying gene explains phenotypic diversity in STXBP1 syndrome
Jovana Kovacevic, PhD
VU Amsterdam

Models of Therapeutic Development

10:25 am ET

Session Chair: Michael Boland, PhD

STXBP1 encephalopathy - mechanisms and therapies interrogated using human brain organoids
Christopher Makinson, PhD
Columbia University

C. elegans and VUS Characterization
Chris Hopkins, PhD
InVivo Biosystems

RNASeq High-throughput Screening
Omid Karkouti, PhD
Rarebase

Next Steps for the STXBP1 Research Network

11:45 am ET

Ben Prosser, PhD
University of Pennsylvania

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