

STXBP1 SUMMIT+

The STXBP1 Summit + Family Meeting will be held virtually 17 - 19 September 2021. All STXBP1 community members are welcome!

Get ready for Research and Clinical updates, Family iSupport and Advocacy sessions, and time to socialize and catch up with your STXBP1 Family.



SCIENCE + LOVE = CURE

Don't Miss These Speakers



Mingshan Xue, PhD
Baylor College of Medicine



Wendy Chung, MD, PhD
Simons Foundation



Ingo Helbig, MD
Children's Hospital of Philadelphia



Matthijs Verhage, PhD
Vrije Universiteit Amsterdam



Cristol Barrett O'Loughlin
Angel Aid



Hannah Stamberger, MD, PhD
University of Antwerp

REGISTRATION:

stxbp1summit.org/register

17 - 19 SEPTEMBER 2021

info@stxbp1disorders.org
www.stxbp1disorders.org

STXBP1 SUMMIT+

Agenda Overview

Friday, 17 September 2021

8:00 pm -
9:30 pm ET

Pajama Dance Party
Featuring Casey Baum

Saturday, 18 September 2021

10:00 am -
2:30 pm ET

Clinical Updates
Translational Research
Regional Lounges

Caregiver Support
Foundation & Advocacy

Sunday, 19 September 2021

10:00 am -
2:30 pm ET

Therapy Development
Natural History Studies
Lounges: Ages, Siblings, Grands, Natural History

Sibling Support
Foundation & Advocacy
Transition to Adulthood

Thanks to Our Sponsors



REGISTRATION:

stxbp1summit.org/register

17 - 19 SEPTEMBER 2021

info@stxbp1disorders.org
www.stxbp1disorders.org

Conference Kickoff

10:00 am ET

Welcome and Kickoff
Charlene Son Rigby
President, STXBPI Foundation

Science

Clinical

10:10 am ET

STXBPI Clinical Update
Ingo Helbig, MD
Children's Hospital of Philadelphia

11:00 am ET

Genetics 101
Sarah McKeown Ruggiero, MS, LCGC
Children's Hospital of Philadelphia

Designing an Approach to Measuring
Development in STXBPI
Ganna Balagura, MD, PhD
University of Genoa

Natural History of STXBPI in
Adulthood
Hannah Stamberger, MD, PhD
University of Antwerp
Elena Gardella, MD, PhD
Danish Epilepsy Centre

Support & Foundation Support

Putting Self back in Self Care -
Caregiver Support
Nikki McIntosh
Rare Mama
Cristol Barrett O'Loughlin
Angel Aid

Translational Research

12:20 pm ET

Mouse and Organoid Models for
Studying STXBPI Disorder
Michael Boland, PhD & Christopher
Makinson, PhD
Columbia University

New Insights in Disease Mechanisms
and New Drug Screening
Matthijs Verhage, PhD & Hannah
Lammertse
Vrije Universiteit Amsterdam
Lukas Neukomm, PhD
University of Lausanne

Foundation and Advocacy

STXBPI Foundation: Where we
started, Where we are, Where we
are going, How You can help
Jennifer Clatterbuck, Melissa Hioco,
Russ Novy, Heather Jones
STXBPI Foundation

1:30 pm ET

Regional Lounges

Catch up with families in your country or region!



Science

Therapy Development

10:00 am ET

Clinical Trial Readiness
Kimberly Goodspeed, MD
University of Texas, Southwestern

Gene Therapy
Mingshan Xue, PhD & Wu Chen, PhD
Baylor College of Medicine, Texas
Children's Hospital

11:10 am ET

Antisense Oligonucleotides (ASOs)
Jennine Dawicki-McKenna, PhD
University of Pennsylvania

Small Molecule Therapies
Jaqueline Burre, PhD
Weill-Cornell Medicine

4-Phenylbutyrate Clinical Trial Update
Zachary Grinspan, MD, MS
Weill-Cornell Medicine

Support & Foundation

Support

Sibling Support Session
STXBP1 Siblings

Recommended 13+ years

Foundation and Advocacy

Advocacy
Jen Lupo Reese
Rare Disease Legislative Advocates

Natural History Studies

12:20 pm ET

Simons Searchlight and STXBP1 Study Update
Wendy Chung, MD, PhD
Simons Foundation

Citizen Natural History Study:
Leveraging technology to increase access to rare disease research
Elli Brimble, MS
Citizen

Support

We ALL need Professional Help - The Transition to Adulthood
Curt Seefeldt & Jennifer A. Disabato,
DNP, CPNP-PC, AC

1:30 pm ET

Lounges

STX'er Age Group, Siblings & Grands Society
Simons Searchlight
Citizen

