The STXB1 Summit + Family Meeting will be held virtually 17 - 19 September 2021. All STXB1 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 STXB1 Family.

Don't Miss These Speakers

Minghsan 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:  
stxb1summit.org/register

17 - 19 SEPTEMBER 2021  
info@stxb1disorders.org  
www.stxb1disorders.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

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Thanks to Our Sponsors

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### Conference Kickoff

**10:00 am ET**
Welcome and Kickoff  
Charlene Son Rigby  
President, STXB1 Foundation

### Science

#### Clinical

**10:10 am ET**  
STXB1 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 STXB1  
  Ganna Balagura, MD, PhD  
  *University of Genoa*

- Natural History of STXB1 in Adulthood  
  Hannah Stamberger, MD, PhD  
  *University of Antwerp*  
  Elena Gardella, MD, PhD  
  *Danish Epilepsy Centre*

### Translational Research

**12:20 pm ET**  
Mouse and Organoid Models for Studying STXB1 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*

### Support & Foundation Support

**11:00 am ET**  
Putting Self back in Self Care - Caregiver Support  
Nikki McIntosh  
*Rare Mama*  
Cristol Barrett O’Loughlin  
*Angel Aid*

### Foundation and Advocacy

**STXB1 Foundation: Where we started, Where we are, Where we are going, How You can help**  
Jennifer Clatterbuck, Melissa Hioco, Russ Novy, Heather Jones  
*STXB1 Foundation*

### 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*

### Natural History Studies

**12:20 pm ET**

Simons Searchlight and STXBPI 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 & Foundation Support

**Sibling Support Session**  
*STXBPI Siblings*  
*Recommended 13+ years*

**Foundation and Advocacy**

Advocacy  
*Jen Lupo Reese*  
*Rare Disease Legislative Advocates*

### Lounges

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