

Join the STXBP1 Summit+ Family Meeting taking place on July 22-23 in Westminster, Colorado!

STXBP1 families will hear from researchers and clinicians who are working to improve the lives of STXBP1 patients and families. Families will also have the opportunity to attend sessions on benefits, advocacy, and learn from other parents.

It is sure to be a weekend full of information and fun... as we learn, connect, and dance the night away at the dinner on Saturday.



STXBP1 Summit+



Scott Demarest, MD
Children's Hospital of Colorado



Allyson Berent, DVM, DACVIM
FAST & Mahzi Therapeutics



Ingo Helbig, MD
Children's Hospital of Philadelphia



Elizabeth Moran, JD
Arc of Colorado



Emma James, PhD
Encoded Therapeutics



Mingshan Xue PhD
Baylor College of Medicine

22 - 23 JULY 2023

Meeting Kickoff & Keynote

7:30 am MT Social Breakfast
Birds of a feather networking

8:30 am MT Charlene Son Rigby, MBA & Melissa Hioco
STXBPI Foundation

Keynote: How Parents Can Drive Drug Development for Rare Disease: From Impossible to Possible

Allyson Berent, DVM, DACVIM
Foundation For Angelman Syndrome Therapeutics (FAST), Mahzi Therapeutics and Angelman Mom

9:30 am MT Clinical

STXBPI Clinical Update

Ingo Helbig, MD & Sarah Ruggiero, MS, LCGC
Children's Hospital of Philadelphia

Genetics 101

Katie Angione, MS, CGC
Children's Hospital of Colorado

Children's Hospital of Colorado's Neurogenetics Clinic

Margarita Saenz, MD & Scott Demarest, MD
Children's Hospital of Colorado

Cortical Visual Impairment in STXBPI-RD

Megan Abbott, MD
Children's Hospital of Colorado

11:25 am MT In the Lab: Translational Research

State of Translational Research in STXBPI, ENDD Center Overview & ASO Development

Michael Boland, PhD
University of Pennsylvania

Progressing AAV gene therapy for STXBPI encephalopathy

Mingshan Xue, PhD
Baylor College of Medicine

STXBP1 SUMMIT+ FAMILY MEETING 2023

Saturday 22 July 2023

12:35 pm MT **Social Lunch**

Birds of a Feather Networking

Featured Family Story

1:35 pm MT **Clinical Trial Readiness & Natural History Studies**

Clinical Trial Readiness

Jennifer Tjernagel, Terry Jo Bichell, PhD & Charlene Son Rigby, MBA
Simons Foundation, COMBINEDBrain, Global Genes & STXBP1 Foundation

Neuropsychological Assessments

Andrea Miele, PhD
Colorado Children's Hospital

ENGAGE Study for STXBP1

Emma James, PhD
Encoded Therapeutics

The ORCA (Observer-Reported Communication Ability) Outcome Measure

Bryce Reeve, PhD
Duke University School of Medicine

Prospective, Multi-site Natural History Study for STXBP1

Katie Rose Sullivan, MS, LCGC
Children's Hospital of Philadelphia

3:40 pm MT **Therapy Development**

Metabolomics study and Biomarker Consortium

James Goss, PhD
STXBP1 Foundation

4-Phenylbutyrate Trial Update

Zachary Grinspan, MD, MS
Weill-Cornell Medicine

6:00 pm MT **Saturday Dinner**

Rockin' in the Rockies Dinner and Dance Party!

6 - 6:30 pm: Pre Dinner Social - *No Host Bar*

6:30 - 9 pm: Social Dinner

STXBPI SUMMIT+ FAMILY MEETING 2023

Sunday 23 July 2023

7:30 am MT **Social Breakfast**

Birds of a Feather Networking
Featured Family Story

8:30 am MT **Be Ready. Be Real. Breakout Session 1**

Caregiver Track Preventing caregiver burnout: A workshop on building resilience

Amelia Seraphia Derr, MSW PhD
Seattle University & STXBPI Mom
Susan Bourgerie, MA
Partners in Resilience & STXBPI Grandma

Advocate Track Justice for All: Leadership in Legislation, Policy, and Practice to Support Our Kids

Elizabeth A. Moran, JD,
Arc of Colorado

10:00 am MT **Be Ready. Be Real. Breakout Session 2**

Caregiver Track Parent to Parent Panel: all things STX and the ups & downs of parenting a child with STXBPI

Moderator: Cristina Brennan, JD
STXBPI Foundation

Advocate Track Benefits for your dependent child/adult including Medicaid & Social Security Disability: Why it's so hard to access benefits and what you can do about it

Spencer Bishins
Licensed attorney & former Social Security Administration (SSA) disability decision writer
Luke Wheeland
The Arc Arapahoe, Douglas & Elbert Counties

11:15 am MT **Closing**

It's a Wrap!
Charlene Son Rigby, MBA & Melissa Hioco
STXBPI Foundation