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

STXBPI families will hear from researchers and clinicians who are working to improve the lives of STXBPI 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.

**Don't Miss These Speakers**

- **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
STXBP1 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

Clinical

9:30 am MT
STXBP1 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 STXBP1-RD
Megan Abbott, MD
Children’s Hospital of Colorado

In the Lab: Translational Research

11:25 am MT
State of Translational Research in STXBP1, ENDD Center Overview & ASO Development
Michael Boland, PhD
University of Pennsylvania

Progressing AAV gene therapy for STXBP1 encephalopathy
Mingshan Xue, PhD
Baylor College of Medicine
Social Lunch

Birds of a Feather Networking

Featured Family Story

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

Therapy Development

Metabolomics study and Biomarker Consortium
James Goss, PhD
STXBP1 Foundation

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

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
STXBP1 SUMMIT+
FAMILY MEETING 2023

Sunday 23 July 2023

7:30 am MT  **Social Breakfast**

Birds of a Feather Networking
Featured Family Story

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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 & STXBP1 Mom*
Susan Bourgerie, MA
*Partners in Resilience & STXBP1 Grandma*

**Advocate Track** Justice for All: Leadership in Legislation, Policy, and Practice to Support Our Kids
Elizabeth A. Moran, JD,
*Arc of Colorado*

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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 STXBP1
Moderator: Cristina Brennan, JD
*STXBP1 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*

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11:15 am MT  **Closing**

It's a Wrap!
Charlene Son Rigby, MBA & Melissa Hioco
*STXBP1 Foundation*