

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*

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**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*

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**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*

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# STXBP1 SUMMIT+ FAMILY MEETING 2023

Saturday 22 July 2023

## 12:35 pm MT **Social Lunch**

Birds of a Feather Networking

Featured Family Story

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## 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*

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## 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*

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## 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

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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 & 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*

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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 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*

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## 11:15 am MT **Closing**

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

# STXBP1 SUMMIT+ FAMILY MEETING 2023



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