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.

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Don't Miss These Speakers

22 - 23 JULY 2023

info@stxbp1disorders.org
www.stxbp1disorders.org

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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

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STXBP1 Summit+
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*

11:25 am MT
In the Lab: Translational Research

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*
临床试验准备及自然历史研究

1:35 pm MT  

临床试验准备
Jennifer Tjernagel, Terry Jo Bichell, PhD & Charlene Son Rigby, MBA  
Simons Foundation, COMBINEDBrain, Global Genes & STXBP1 Foundation

神经心理学评估
Andrea Miele, PhD  
Colorado Children's Hospital

ENGAGE研究STXBP1
Emma James, PhD  
Encoded Therapeutics

ORCA（观察报告沟通能力）结果测量
Bryce Reeve, PhD  
Duke University School of Medicine

多中心自然历史研究STXBP1
Katie Rose Sullivan, MS, LCGC  
Children's Hospital of Philadelphia

治疗发展

3:40 pm MT  

代谢组学研究和生物标志物合作
James Goss, PhD  
STXBP1 Foundation

4-苯基丁酸盐试验更新
Zachary Grinspan, MD, MS  
Weill-Cornell Medicine

周六晚餐

6:00 pm MT  

Rockin' in the Rockies Dinner and Dance Party!

6 - 6:30 pm: Pre Dinner Social - No Host Bar  
6:30 - 9 pm: Social Dinner
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*

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*

11:15 am MT  **Closing**  
It's a Wrap!  
Charlene Son Rigby, MBA & Melissa Hioco  
*STXBP1 Foundation*