



STXBP1
Global Connect



 **STXBP1**
disorders
Science + Love = Cure



2nd European STXBP1 | October 8-11, 2025
Summit and Research Roundtable | Heidelberg, Germany

October 8-10, 2025
Research Summit

Center for internal medicine (Kreihl Klinik)
Im Neuenheimer Feld 410
69120 Heidelberg

October 11, 2025
Family Summit

Center for pediatrics
Im Neuenheimer Feld 430
69120 Heidelberg

Organization Committee

Steffen Syrbe
Kim Marie Thalwitzer
Ganna Balagura
Hannah Stamberger
Matthijs Verhage
Pasquale Striano
Fabienne Pietrusky
Katja Hoffmann

stxbp1eu.org



RESEARCH SUMMIT DAY 1 | WEDNESDAY 8th

- 14.00** Registrations
- 14.30** Kick-Off **Steffen Syrbe (Heidelberg)**
- 14.45** **Session 1: Shared and distinct mechanisms for monogenic neurodevelopmental disorders**
- Introduction to SNAREopathies **Matthijs Verhage (Amsterdam)**
SNAREopathies; the SYT1 case **Niels Cornelisse (Amsterdam)**
Inborn errors of synaptic transmission **Thomas Opladen (Heidelberg)**
Autophagy in epileptic disorders from SNAP-SNARE-synaptopathies **Afshin Saffari (Heidelberg)**
Hot topic emerging in 2025
- 16.00** *Coffee break*
- 16.15** **Session 2: Disease mechanisms of STXBP1**
- New disease mechanisms for STXBP1 missense mutations **Sylvia Korhorn (Amsterdam)**
Syntaxin Dysfunctions **Jacqueline Burre (New York)**
In vitro modeling of Patient-Specific STXBP1 Variants **Caroline Pearson (New York)**
STXBP1 haploinsufficiency and homeostatic responses at the cellular levels **Romina Ambrosini Defendi (Amsterdam)**
Microcircuit failure in STXBP1 encephalopathy leads to hyperexcitability **Jean-Francois Perrier (Copenhagen)**
- 17.45** **Expert discussion**
- 18.15** **Conference reception**



RESEARCH SUMMIT DAY 2 | THURSDAY 9th

9.00 **Session 3: Clinical science on STXBP1-related disorders**

Gait control study for STXBP1 related disorders **Hannah Stamberger (Antwerpen)**
Mild cases of STXBP1 **Francesca Furia (Dianalund)**
MRI study of STXBP1-related disorders **Matteo Lenge/Alice Dainelli (Florence)**
Twin study of STXBP1-related disorders **Kim Thalwitzer (Heidelberg)**
Hot topic emerging in 2025

10.15 *Coffee break*

10.30 **Session 4: Standard of care of STXBP1-related disorders**

New insights into epileptic encephalopathies **Steffen Syrbe (Heidelberg)**
Clinical update on STXBP1-related disorders **Ingo Helbig (Philadelphia)**
Delphi protocol for STXBP1 **Ganna Balagura (Genoa)**
Hot topic emerging in 2025
Hot topic emerging in 2025

11.30 **Expert discussion**

12.15 *Lunch break & poster session*

13.45 **Session 5: Therapeutic development I**

Rafa's Moonshot and the potential of NMN **Hila Ben-Moshe (Tel Aviv)**
ASO strategies to treat STXBP1 haploinsufficiency **Ben Prosser (Philadelphia)**
CRISPRa strategies to treat STXBP1 haploinsufficiency **Nathan Hendersen (Philadelphia)**
Genetic Therapy Development in mice **Mingshan Xue (Houston)**
Disease-Modifying Treatment for STXBP1: rAAV CAP-002 **Capsida Biotherapeutics**

15.00 *Coffee break*

15.15 **Session 6: Therapeutic development II**

Natural History Study and Gene Therapy for *DRAVET* syndrome **Andreas Brunklaus (Glasgow)**
Implementation of gene therapy – lessons learned from neuromuscular diseases **Andreas Ziegler (Heidelberg)**
Hot topic emerging in 2025
Hot topic emerging in 2025
Hot topic emerging in 2025

16.30 **Expert Discussion**

19.00 **Conference dinner**



RESEARCH SUMMIT DAY 3 | FRIDAY 10th

9.00 **Session 7: Biomarker and endpoint development**

EEG-biomarker studies **Hilgo Bruining (Amsterdam)**

Mapping the resting state EEG functional connectome in STXBP1 syndrome **Additya Sharma (Amsterdam)**

qEEG based quantification of developmental consequences in STXBP1 syndrome **Shilpa Anand (Amsterdam)**

Quantitative EEG biomarkers of severity in STXBP1-related disorders **Jillian McKee (Philadelphia)**

Smartphone applications for seizure diaries **ESCO**

Multiomics biomarkers in STXBP1 **Ganna Balagura (Genoa)**

10.15 *Coffee break*

10.30 **Session 8: Clinical trial readiness**

The STARR natural history study **Sarah Ruggiero (Philadelphia)**

The ESCO registry **Elena Gardella (Dianalund)**

The ESCO natural history study **Hannah Stamberger (Antwerpen)**

A consensus Clinical Global Impression (GCI) scale for STXBP1 Disorders **Sarah Tefft (Philadelphia)**

Pilot and Validation of a novel STXBP1-Clinical Severity Assessment **Megan Abbott (Colorado)**

The best version of the Bayley 4 for STXBP1: an item analysis **Andrea Miele (Colorado)**

11.45 **Expert discussion**

Faculty:

Matthijs Verhage (Amsterdam), Niels Cornelisse (Amsterdam), Thomas Opladen (Heidelberg), Afshin Saffari (Heidelberg), Sylvia Korhorn (Amsterdam), Jacqueline Burre (New York), Caroline Pearson (New York), Romina Ambrosini Defendi (Amsterdam), Jean-Francois Perrier (Copenhagen), Hannah Stamberger (Antwerpen), Francesca Furia (Dianalund), Matteo Lenge (Florence), Alice Dainelli (Florence), Kim Thalwitzer (Heidelberg), Ingo Helbig (Philadelphia), Ganna Balagura (Genoa), Hila Ben-Moshe (Tel Aviv), Ben Prosser (Philadelphia), Nathan Hendersen (Philadelphia), Mingshan Xue (Houston), Andreas Brunklaus (Glasgow), Andreas Ziegler (Heidelberg), Hilgo Bruining (Amsterdam), Jillian McKee (Philadelphia), Additya Sharma (Amsterdam), Shilpa Anand (Amsterdam), Sarah Ruggiero (Philadelphia), Elena Gardella (Dianalund), Sarah Tefft (Philadelphia), Megan Abbott (Colorado), Andrea Miele (Colorado), Hannah Stamberger (Antwerpen), Fabienne Pietrusky (STXBP1 e.V.), Katja Hoffmann (STXBP1 e.V.), Charlene Son Rigby (STXBP1 Foundation), Mathieu Milh (Marseille), Bruria Ben Zeev (Ramat Gan), Sarah Weckhuysen (Antwerpen), Francesca Furia (Philadelphia), Alejandra Darling (Barcelona), Mila Misra (Amsterdam), Steffen Syrbe (Heidelberg), Capsida Biotherapeutics



FAMILY DAY – SCIENTIFIC PROGRAM | SATURDAY 11th

- 8.30 Registration
- 9.00 Kick Off (**German family group**) & Family story (**Fabienne Pietrusky & Katja Hoffmann**)
- 9.15 **STXBP1 in a short run**
- Genetics & pathophysiology **Hannah Stamberger (Antwerpen) & Matthijs Verhage (Amsterdam)**
Clinical presentation & standard of care **Steffen Syrbe (Heidelberg)**
FAQ
- 9.45 **Presentation of family groups**
- 10.45 *Coffee break & get together*
- 11.15 **Sumup of the research summit 2025** **Kim Thalwitzer (Heidelberg)**
- 11.35 **Session 2: Ongoing studies**
- ESCO natural history study & registry **Ganna Balagura (Genoa)**
STARR natural history study **Sarah Ruggiero (Philadelphia)**
FAQ
- 12.20 *Lunch break*
- 14.20 **Roundtable 1: What does it mean to participate in a therapeutic trial?**
- 15.35 *Coffee break*
- 15.50 **Roundtable 2: Unmet needs & support of patients & families**
- 17.05 **Q & A**



ACTIVITIES AT THE FAMILY SUMMIT

- Live streaming rooms
- Playroom
- Possibility to participate in research projects
- 1 on 1 genetic counseling sessions (Katie Angione, Colorado & Sarah Ruggiero, Phialdelphia)

