



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 (Krehl 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

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RESEARCH SUMMIT DAY 1 | WEDNESDAY 8th

13.00 *Registrations*

14.20 **Kick-Off** Steffen Syrbe (Heidelberg)

14.25 *European STXBP1 association*

14.45 **Session 1: Shared and distinct mechanisms for monogenic neurodevelopmental disorders**

Chair: Matthijs Verhage (Amsterdam)

Matthijs Verhage (Amsterdam) Introduction to SNAREopathies

Niels Cornelisse (Amsterdam) SNAREopathies: the SYT1 case

Jacqueline Burre (New York) Syntaxin Dysfunctions (*online*)

Thomas Opladen (Heidelberg) Inborn errors of synaptic transmission

15.50 *Coffee break*

16.20 **Session 2: Disease mechanisms of STXBP1-related disorders**

Chair: Ruud Toonen (Amsterdam)

Sylvia Korhorn (Amsterdam) Functional analysis of STXBP1 missense mutations discriminates three disease mechanisms

Caroline Pearson (New York) In vitro modeling of Patient-Specific STXBP1 Variants (*online*)

Romina Ambrosini Defendi (Amsterdam) STXBP1 haploinsufficiency and homeostatic responses at the cellular levels

Jean-Francois Perrier (Copenhagen) Microcircuit failure in STXBP1 encephalopathy leads to hyperexcitability

Matthew Vanheusden (Amsterdam) Early developmental effects of STXBP1 deficiency

17.40 **Expert discussion** *Chair: Matthijs Verhage (Amsterdam)*

18.30 **Conference reception**



RESEARCH SUMMIT DAY 2 | THURSDAY 9th

9.00 Session 3: Clinical science on STXBP1-related disorders *Chair: Kim Thalwitzer (Heidelberg)*

Ingo Helbig (Philadelphia)	Clinical update on STXBP1-related disorders
Merel Swinnen (Antwerpen)	Gait control study for STXBP1 related disorders
Francesca Furia (Dianalund)	Mild cases of STXBP1 related disorders
Adrián Valls Carbó (Madrid)	STXBP1 cohort description
Matteo Lenge/Alice Dainelli (Florence)	MRI study of STXBP1-related disorders

10.20 *Coffee break*

10.50 Session 4: Therapeutic development - lessons from other neurodevelopmental disorders

Chair: Bruria Ben-Zeev (Ramat Gan)

Bruria Ben-Zeev (Ramat Gan)	Introduction to precisional therapies in DEEs
Allan Bayat (Dianalund)	Recent advances in treating DEEs – lessons from Dravet & Lennox Gestaut
Matias Wagner (Munich)	From innovative diagnostics to personalized treatments - lessons from ASO development in SCN2A
Andreas Ziegler (Heidelberg)	Implementation of gene therapy – lessons learned from neuromuscular diseases

12.00 *Lunch break & poster session*

13.20 Session 5: Therapy approaches for STXBP1-related disorders I *Chair: Ganna Balagura (Genoa)*

Ana Carreras Mascaro (Neurospecter)	Therapy development in human neurons in vitro
Efrat Ben-Zeev (Weizmann Institute of Science, Rehovot)	Advancing STXBP1 Research: Structural Insights and Computational Approaches for Small Molecule Discovery
Millie Stone (New York)	Updates from the Ravicti Study
Hila Ben-Moshe (Tel Aviv)	Rafa's Moonshot and the potential of NMN

14.25 *Group photo + Coffee break*

15.00 Session 6: Therapy approaches for STXBP1-related disorders II *Chair: James Goss (STXBP1 foundation)*

Mingshan Xue (Houston)	Therapy development in mice
Ben Prosser (Philadelphia)	ASO strategies to treat STXBP1 haploinsufficiency
Gad Vatine (Beer Sheva)	Targeted RNA editing as a potential treatment for G>A Mutations in STXBP1-Related Disorders
Nathan Henderson (Philadelphia)	CRISPRa strategies to treat STXBP1 haploinsufficiency

16.00 Roundtable 1– Promoting academia-industry partnership *Chair: Claudia Persoon (Neurospecter)*

19.00 *Conference dinner*



RESEARCH SUMMIT DAY 3 | FRIDAY 10th

9.00

Session 7: Biomarker and endpoint development

Chair: Elena Gardella (Dianalund)

Hilgo Bruining (Amsterdam)

EEG-biomarker studies

Additya Sharma (Amsterdam)

Mapping the resting state EEG functional connectome in STXBP1 syndrome

Shilpa Anand (Amsterdam)

qEEG based quantification of developmental consequences in STXBP1 syndrome

Jillian McKee (Philadelphia)

Quantitative EEG biomarkers of severity in STXBP1-related disorders

Laura Wetzel/Jan Lui (BioMarin)

Plasma biomarkers for STXBP1-related disorders

10.10

Coffee break

10.40

Session 8: Clinical trial readiness

Andreas Brunklaus (Glasgow)

Natural History Study and Gene Therapy for *DRAVET* syndrome

Samuel Pierce (Philadelphia)

The STARR natural history study

Elena Gardella (Dianalund)

The ESCO registry

Hannah Stamberger (Antwerpen)

The ESCO natural history study

Sarah Tefft (Philadelphia)

A consensus Clinical Global Impression (GCI) scale for STXBP1 Disorders

Andrea Miele (Colorado)

The best version of the Bayley 4 for STXBP1: an item analysis

Megan Abbott (Colorado)

Pilot and Validation of a novel STXBP1-Clinical Severity Assessment

12.10

Expert discussion



FAMILY DAY | SATURDAY 11th

- 8.30** Registration
- 9.00** Kick Off (**German family group**) & Family story (**Fabienne Pietrusky & Katja Hoffmann**)
- 9.15** **Session 1: STXBP1 in a short run**
- Genetics & disease mechanisms **Hannah Stamberger (Antwerpen) & Matthijs Verhage (Amsterdam)**
- Clinical presentation & standard of care **Ingo Helbig (Philadelphia)**
- FAQ
- 9.45** **Presentation of family groups**
- 10.45** *Coffee break*
- 11.15** **Sumup of the research summit 2025** **Kim Thalwitzer (Heidelberg)**
- 11.35** **Session 2: Ongoing studies**
- ESCO natural history study & registry **Ganna Balagura (Genua)**
- STARR natural history study **Ingo Helbig (Philadelphia)**
- FAQ
- 12.20** *Lunch break*
- 14.20** **Global SUDEP Update: Research and Progress** **Oskar Killinger Stiftung**
- 14.30** **Roundtable 1: Therapies in Development for STXBP1 and Clinical Trials**
- Participants:*
- James Goss (**STXBP1** Foundation)
- Charlene Son Rigby (**STXBP1** Foundation)
- Hilgo Bruining (Amsterdam)
- Scott Damarest (Colorado)
- 15.45** *Coffee break*
- 16.15** **Roundtable 2: Unmet needs, organization & support of patients & families**
- Participants:*
- Kelsey Ax (Amsterdam)
- Sietske van Till (Rotterdam)
- Gilberte Schnur (**STXBP1** Germany)