



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
Fabienne Pietrusky
Katja Hoffmann

stxbp1eu.org



RESEARCH SUMMIT DAY 1 | WEDNESDAY 8th

	Name	Title	Duration Talk + Questions
13.00	<i>Registrations</i>		
14.20	Kick-Off Steffen Syrbe (Heidelberg)		
14.30	Session 1: Shared and distinct mechanisms for monogenic neurodevelopmental disorders		
	<i>Chair: Matthijs Verhage (Amsterdam)</i>		
	Matthijs Verhage (Amsterdam)	Introduction to SNAREopathies	13 + 2
	Niels Cornelisse (Amsterdam)	SNAREopathies: the SYT1 case	13 + 2
	Jacqueline Burre (New York)	Syntaxin Dysfunctions	13 + 2 (online)
	Thomas Opladen (Heidelberg)	Inborn errors of synaptic transmission	13 + 2
15.50	<i>Coffee break</i>		
	Session 2: Disease mechanisms of STXBP1-related disorders		
16.20	<i>Chair: Ruud Toonen (Amsterdam)</i>		
	Sylvia Kothorn (Amsterdam)	Functional analysis of STXBP1 missense mutations discriminates three disease mechanisms	13 + 2
	Caroline Pearson (New York)	In vitro modeling of Patient-Specific STXBP1 Variants	13 + 2 (online)
	Romina Ambrosini Defendi (Amsterdam)	STXBP1 haploinsufficiency and homeostatic responses at the cellular levels	13 + 2
	Jean-Francois Perrier (Copenhagen)	Microcircuit failure in STXBP1 encephalopathy leads to hyperexcitability	13 + 2
	Matthew Vanheusden (Amsterdam)	Early developmental effects of STXBP1 deficiency	13 + 2
17.40	Expert discussion <i>Chair: Matthijs Verhage (Amsterdam)</i>		
18.30	Conference reception		



RESEARCH SUMMIT DAY 2 | THURSDAY 9th

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

Hannah Stamberger (Antwerpen)	Gait control study for STXBP1 related disorders	13 + 2
Francesca Furia (Dianalund)	Mild cases of STXBP1 related disorders	13 + 2
Antonio Gil Nagel team (Madrid)	Longitudinal study on STXBP1	13 + 2
Matteo Lenge/Alice Dainelli (Florence)	MRI study of STXBP1-related disorders	13 + 2
Helbig Group	Clinical update on STXBP1-related disorders	13 + 2

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	5
Allan Bayat (Dianalund)	Recent advances in treating DEEs – lessons from Dravet & Lennox Gestaut	13 + 2
Matias Wagner (Munich)	From innovative diagnostics to personalized treatments - lessons from ASO development in SCN2A	13 + 2
Andreas Ziegler (Heidelberg)	Implementation of gene therapy – lessons learned from neuromuscular diseases	13 + 2

12.00 *Lunch break & poster session*

13.20 Session 5: Standard of care and current therapies for STXBP-related disorders *Chair: Ganna Balagura*

Ganna Balagura (Genoa)	Introduction & Delphi protocol for STXBP1	13 + 2
Millie Stone (New York)	Updates from the Ravict Study	13 + 2
Capsida Biotherapeutics	Disease-Modifying Treatment for STXBP1: rAAV CAP-002	13 + 2
Hila Ben-Moshe (Tel Aviv)	Rafa's Moonshot and the potential of NMN	13 + 2

14.25 *Group photo + Coffee break*

15.00 Session 6: New therapy development for STXBP1-related disorders *Chair: James Goss*

Ana Carreras Mascaro (Neurospector)	Therapy development in human neurons in vitro	13 + 2
Mingshan Xue (Houston)	Therapy development in mice	13 + 2
Ben Prosser (Philadelphia)	ASO strategies to treat STXBP1 haploinsufficiency	13 + 2
Gad Vatine (Beer Sheva)	Targeted RNA editing as a potential treatment for G>A Mutations in STXBP1-Related Disorders	13 + 2
Nathan Henderson (Philadelphia)	CRISPRa strategies to treat STXBP1 haploinsufficiency	13 + 2
Efrat Ben-Zeev (Rehovot)	Advancing STXBP1 Research: Structural Insights and Computational Approaches for Small Molecule Discovery	13 + 2

16.40 Roundtable 1– Promoting academia-industry partnership *Chair: Claudia Persoon (Neurospector)*

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	13 + 2
Additya Sharma (Amsterdam)	Mapping the resting state EEG functional connectome in STXBP1 syndrome	8 + 2
Shilpa Anand (Amsterdam)	qEEG based quantification of developmental consequences in STXBP1 syndrome	8 + 2
Jillian McKee (Philadelphia)	Quantitative EEG biomarkers of severity in STXBP1-related disorders	13 + 2
Laura Wetzel/Jan Lui (BioMarin)	Plasma biomarkers for STXBP1-related disorders	13 + 2

10.10

Coffee break

10.40

Session 8: Clinical trial readiness

Andreas Brunklaus (Glasgow)	Natural History Study and Gene Therapy for <i>DRAVET</i> syndrome	13 + 2
Sarah Ruggiero (Philadelphia)	The STARR natural history study	13 + 2
Elena Gardella (Dianalund)	The ESCO registry	8 + 2
Hannah Stamberger (Antwerpen)	The ESCO natural history study	8 + 2
Sarah Tefft (Philadelphia)	A consensus Clinical Global Impression (GCI) scale for STXBP1 Disorders	8 + 2
Megan Abbott (Colorado)	Pilot and Validation of a novel STXBP1-Clinical Severity Assessment	8 + 2
Andrea Miele (Colorado)	The best version of the Bayley 4 for STXBP1: an item analysis	8 + 2

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** **STXBP1 in a short run**
- Genetics & disease mechanisms **Hannah Stamberger (Antwerpen) & Matthijs Verhage (Amsterdam)**
- Clinical presentation & standard of care **Helbig group (Philadelphia)**
- 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 (Genua)**
- STARR natural history study **Sarah Ruggiero (Philadelphia)**
- FAQ
- 12.20** *Lunch break*
- 14.20** **Roundtable 2: What does it mean to participate in a therapeutic trial?**
- Participants:*
- James Goss (*STXBP1* Foundation)
- Charlene Son Rigby (*STXBP1* Foundation)
- Hilgo Bruining (Amsterdam)
- 15.35** *Coffee break*
- 15.50** **Roundtable 3: Unmet needs, organization & support of patients & families**
- Participants:*
- Kelsey Ax (Amsterdam)
- Sietske van Till (Rotterdam)
- European Families
- 17.05** **Q & A**