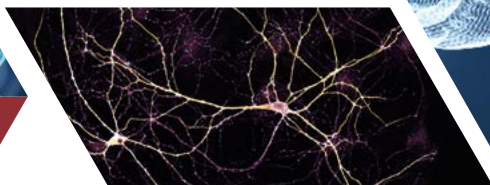




This Networking event has received funding from the European Union's Horizon 2020 research and innovation programme under the EJP RD COFUND-EJP N° 825575

# 1<sup>st</sup> European STXBP1 Summit and Research Roundtable

**16-18 May 2023**  
**MILAN (Italy)**



Organization Committee  
**Ganna Balagura**  
**Charlene Son Rigby**  
**Matthijs Verhage**  
**Sarah Weckhuysen**

## DAY 1 | TUESDAY 16<sup>TH</sup>

### RESEARCH MEETING: STXBP1 IN THE LAB

**8.30** Registrations

**9.00** Opening and Introduction **G. Balagura**

### SESSION I

#### Mechanisms and models of disease in STXBP1- related disorders

Chairs: **M. Verhage & J. Goss**

**9.10** A history of STXBP1 research: where are we now **M. Verhage**

**9.25** Single cell transcriptional changes in an STXBP1 mouse model **A. Roig Adam**

**9.45** Microcircuit failure in STXBP1 encephalopathy leads to hyperexcitability, rescue with AMPAkinases **A. Britos dos Santos, J. F. Perrier**

**10.10** Disease-linked mutations in Munc18-1 deplete synaptic proteins **N. Guiberson**

**10.30** *Coffee Break*

**11.00** Synaptic phenotypes in patient-derived neurons **J. Sørensen**

**11.20** Network defects and proteome changes in patient-derived neurons **M. Öttl**

**11.40** Modelling STXBP1 encephalopathy in human brain slice cultures **F. McLeod**

**12.00** Early Network Activity in a mouse model of STXBP1 disorder **P. P. Lenck-Santini**

**12.20** *Lunch*

### SESSION II

#### Pipeline towards therapies for STXBP1- related disorders

Chair: **F. Zara**

**14.00** Fly screen with small molecules **L. Neukomm, C. Schäfer**

**14.20** ASOs and miRNA modulation for STXBP1-RD **B. Prosser**

**14.40** ASOs to inhibit poison exons in STXBP1 **L. Janssen**

**14.55** CRISPR-editing as potential therapeutic strategy **M. Boland**

**15.15** *Coffee Break*

**15.45** Gene replacement therapy design **R. Toonen**

**16.05** A preclinical pharmacology study of AAV gene therapy in a mouse model of STXBP1 encephalopathy **M. Xue**

**16.25** Cell-based therapy with GABAergic precursors for STXBP1-RD **M. Álvarez Dolado**

**17.00** **Roundtable of pre-clinical researchers**

Chair: **M. Verhage**

**18.00** Closing remarks

## DAY 2 | WEDNESDAY 17<sup>TH</sup>

### RESEARCH MEETING: STXBP1 IN THE CLINIC

#### SESSION III

##### The clinical spectrum and standard of care in STXBP1-related disorders

Chairs: **A. Garcia Cazorla & M. Milh**

- 9.00** Phenotypic and genotypic spectrum of STXBP1-RD: updates **I. Helbig**
- 9.20** Multilevel phenotyping of STXBP1 patients: the N=You approach **H. Bruining**
- 9.40** Sensory and behavioral profiles in people with STXBP1-RD **A. Fetta**
- 10.00** Survey with STXBP1 families **C. Son Rigby**
- 10.20** Cortical Visual Impairment in STXBP1-RD **M. Abbott**
- 10.40** Launch of the European STXBP1 registry **F. Furia, G. Balagura, H. Stamberger, E. Gardella**
- 11.00** *Coffee Break*
- 11.20** Anti-seizure medications in STXBP1-RD and role of NETRE **S. Syrbe**
- 11.40** The role of 4-Phenyl-Butyrate in STXBP1-RD: clinical trial updates **Z. Grinspan**
- 12.00** Integrative therapeutic approaches and evidence-based rehabilitation strategies in NDDs  
**A. Guzzetta**
- 12.20** Childrens' Colorado model of a Neurogenetic Multidisciplinary Clinic **S. Demarest**
- 12.40** Neuropsychological assessment in children with STXBP1-RD **A. Miele**
- 13.00** *Lunch*

#### SESSION IV

##### The future therapeutic landscape in STXBP1-related disorders: what to treat, how to measure?

Chairs: **G. Balagura & P. Striano**

- 14.00** Introduction to the session: Targeted therapies for STXBP1-RD **J. Goss**
- 14.20** Clinical trials for rare disorders: challenges and novel strategies **S. Auvin**
- 14.40** EEG in STXBP1-RD: not only epilepsy **E. Gardella**
- 15.00** qEEG analysis in STXBP1-disorders **K. Linkenkaer-Hansen**
- 15.20** Characterizing gait deviations in DEE and potential impact for rehabilitation **A. Hallemans**
- 15.40** Gut-brain axis in epilepsy and autism **E. Russo**
- 16.00** *Coffee Break*
- 16.20** NHS Natural history **S. Ruggiero**
- 16.40** ESCO natural history study **G. Balagura, M. Verhage**
- 17.00** Angelman Biomarker Outcome Consortium **J. Panagoulas**
- 17.20** The model of CDKL5 consortium **X. Liogier**
- 17.40** Roundtable of clinical researchers
- 18.40** Closing remarks

**MILAN May 2023**

## DAY 3 | THURSDAY 18<sup>TH</sup>

FAMILIES & RESEARCHER MEETING: STXBP1 AT HOME

### SESSION V

STXBP1 family community development and integration (hybrid session)

Chair: **C. Son Rigby**

**9.00** Family community development and integration **C. Son Rigby**

**9.15** Overview of progress and state of the art (day 1+2)

**G. Balagura & S. Weckhuysen**

**9.45** **Regional STXBP1 associations: introductions & goals (hybrid session)**

**11.00** *Coffee Break*

**11.30** **Roundtable of PAOs, health-care professionals and researchers**

Chair: **C. Son Rigby**

**13.00** *Lunch*

**14.00** ESCO consortium meeting (closed session)

**18.00** Closing remarks

## FACULTY

Megan **Abbott**, *Norway (USA)*  
Manuel **Álvarez Dolado**, *Sevilla (E)*  
Stephane **Auvin**, *Paris (F)*  
Ganna **Balagura**, *Genoa (IT)*  
Michael **Boland**, *New York (USA)*  
Altair **Britos dos Santos**, *Copenhagen (DK)*  
Hilgo **Bruining**, *Amsterdam (NL)*  
Scott **Demarest**, *Aurora (USA)*  
Anna **Fetta**, *Bologna (IT)*  
Francesca **Furia**, *Odense (DK)*  
Angeles **Garcia Cazorla**, *Barcelona (E)*  
Elena **Gardella**, *Odense (DK)*  
James **Goss**, *Holly Springs (USA)*  
Zachary **Grinspan**, *New York (USA)*  
Noah **Guiberson**, *New York (USA)*  
Andrea **Guzzetta**, *Pisa (IT)*  
Anne **Hallems**, *Antwerp (B)*  
Ingo **Helbig**, *Philadelphia (USA)*  
Lara **Janssen**, *Amsterdam (NL)*  
Pierre-Pascal **Lenck-Santini**, *Marseille (F)*  
Klaus **Linkenkaer-Hansen**, *Amsterdam (NL)*  
Xavier **Liogier**, *London (UK)*  
Faye **McLeod**, *Newcastle (UK)*  
Andrea **Miele**, *Aurora (USA)*  
Mathieu **Milh**, *Marseille (F)*  
Lukas **Neukomm**, *Lausanne (CH)*  
Miriam **Öttl**, *Amsterdam (NL)*  
Jennifer **Panagoulis**, *Austin (USA)*  
Jean-Francois **Perrier**, *Copenhagen (DK)*  
Ben **Prosser**, *Philadelphia (USA)*  
Amparo **Roig Adam**, *Amsterdam (NL)*  
Sarah **Ruggiero**, *Philadelphia (USA)*  
Emilio **Russo**, *Catanzaro (IT)*  
Christel **Schäfer**, *Lausanne (CH)*  
Charlene **Son Rigby**, *San Francisco (USA)*  
Jakob **Sørensen**, *Copenhagen (DK)*  
Hannah **Stamberger**, *Antwerp (B)*  
Pasquale **Striano**, *Genoa (IT)*  
Steffen **Syrbe**, *Heidelberg (DEU)*  
Ruud **Toonen**, *Amsterdam (NL)*  
Matthijs **Verhage**, *Amsterdam (NL)*  
Sarah **Weckhuysen**, *Antwerp (B)*  
Mingshan **Xue**, *Houston (USA)*

## ORGANIZATION COMMITTEE

Ganna **Balagura**  
*IRCCS G. Gaslini Institute, Italy*  
Charlene **Son Rigby**  
*STXBP1 Foundation, USA*  
Matthijs **Verhage**  
*VU Amsterdam, The Netherlands*  
Sarah **Weckhuysen**  
*University of Antwerp, Belgium*

## CONGRESS VENUE

**Hotel NH Milano Touring**  
Via Iginio Ugo Tarchetti, 2  
20121 Milano

## ORGANIZING SECRETARIAT

**SYMPOSIA** | We care  
ORGANIZZAZIONE CONGRESSI

Symposia Organizzazione Congressi Srl  
Provider Standard Age.Na.S. n. 486  
Piazza Campetto, 2/8 - 16123 Genova  
**T**el. 010 255146

**E** symposia@symposiacongressi.com  
**W** www.symposiacongressi.com

**MILAN May 2023**

**1<sup>st</sup> European STXBP1 Summit  
and Research Roundtable**

