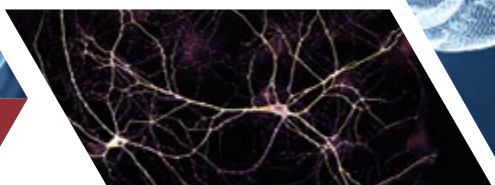


# **1<sup>st</sup> European STXBP1 Summit and Research Roundtable**

**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** Registration  
**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-RD **P. P. Lenck-Santini**  
**12.20** qEEG analysis in STXBP1-RD **K. Linkenkaer-Hansen**  
**12.40** *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** Uncovering the biology of STXBP1 poison exons **L. Janssen**  
**14.55** CRISPR-mediated transcriptional activation 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 preclinical 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** Multilevel phenotyping of STXBP1 patients: the N=You approach **H. Bruining**
- 9.20** Sensory and behavioral profiles in people with STXBP1-RD **A. Fetta**
- 9.40** STXBP1 ENGAGE Study: Capturing Patient and Family Lived Experiences **J. Gofshteyn**
- 10.00** Childrens' Colorado model of a Neurogenetic Multidisciplinary Clinic **S. Demarest**
- 10.20** Launch of the European STXBP1 registry **F. Furia, G. Balagura, H. Stamberger, E. Gardella**
- 10.40** *Coffee Break*
- 11.00** Role of Anti-Seizure Medications in STXBP1-RD – Relevance and help from clinical Networking **S. Syrbe**
- 11.20** The role of 4-Phenyl-Butyrate in STXBP1-RD: clinical trial updates **Z. Grinspan**
- 11.40** Standardized assessments in physical and occupational therapy in STXBP1-RD **S. Pierce, K. V. Cunningham**
- 12.00** Cortical Visual Impairment in STXBP1-RD **M. Abbott**
- 12.20** The emerging scope of neuropsychological abilities in STXBP1 **A. Miele**
- 12.40** *Lunch*

#### SESSION IV

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

Chairs: **P. Striano & S. Weckhuysen**

- 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**
- 15.20** 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** Prospective, Multi-Site Natural History Study of STXBP1-RD **S. Ruggiero**
- 16.40** ESCO natural history study **G. Balagura, M. Verhage**
- 17.00** Angleman Biomarker Outcome Consortium **J. Panagoulas**
- 17.20** The Loulou Foundation CDKL5 pre-competitive consortium **X. Liogier**
- 17.40** Virtual clinical trials and seizure prediction in STXBP1-RD **J. Xian**
- 18.00** Roundtable of clinical researchers and industry
- 19.00** Closing remarks

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

FAMILIES & RESEARCHERS 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 & H. Stamberger**
- 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** Roundtable of STXBP1 families 2
- 15.30** Closing remarks
- 16.00-19.00** ESCO consortium meeting (closed session)

## FACULTY

Megan **Abbott**, *Aurora (USA)*  
Manuel **Álvarez Dolado**, *Sevilla (E)*  
Stephane **Auvin**, *Paris (F)*  
Ganna **Balagura**, *Genova (IT)*  
Michael **Boland**, *New York (USA)*  
Altair **Britos dos Santos**, *Copenhagen (DK)*  
Hilgo **Bruining**, *Amsterdam (NL)*  
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*STXBP1 Foundation, USA*  
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*VU Amsterdam, The Netherlands*  
Sarah **Weckhuysen**  
*University of Antwerp, Belgium*

## CONGRESS VENUE

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

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**MILAN 2023**

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