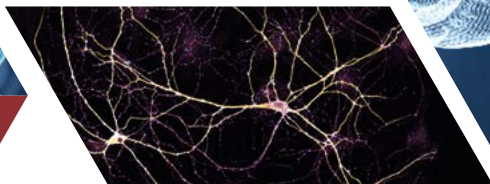




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1st European STXBP1 Summit and Research Roundtable

16-18 May 2023
MILAN (Italy)



Organization Committee
Ganna Balagura
Charlene Son Rigby
Matthijs Verhage
Sarah Weckhuysen

1st European STXBP1 Summit and Research Roundtable

DAY 1 | TUESDAY 16TH

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 Uncovering the biology of STXBP1 poison exons **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 17TH

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** STXBP1 ENGAGE Study: Capturing Patient and Family Lived Experiences **J. Gofshteyn**
- 10.20** Children's Colorado model of a Neurogenetic Multidisciplinary Clinic **S. Demarest**
- 10.40** Launch of the European STXBP1 registry **F. Furla, G. Balagura, H. Stamberger, E. Gardella**
- 11.00** *Coffee Break*
- 11.20** Role of Anti-Seizure Medications in STXBP1 -RD – Relevance and help from clinical Networking **S. Syrbe**
- 11.40** The role of 4-Phenyl-Butyrate in STXBP1-RD: clinical trial updates **Z. Grinspan**
- 12.00** Standardized assessments in physical and occupational therapy in STXBP1-related disorders **S. Pierce, K. V. Cunningham**
- 12.20** Cortical Visual Impairment in STXBP1-RD **M. Abbott**
- 12.40** The emerging scope of neuropsychological abilities in STXBP1 **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** Prospective, Multi-Site Natural History Study of STXBP1-related Disorder **S. Ruggiero**
- 16.40** ESCO natural history study **G. Balagura, M. Verhage**
- 17.00** Angelman 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-related disorders **J. Xian**
- 18.00** Roundtable of clinical researchers and industry
- 19.00** Closing remarks

DAY 3 | THURSDAY 18TH

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** Roundtable of STXBP1 families 2
- 15.30** Closing remarks
- 16.00-19.00** ESCO consortium meeting (closed session)

FACULTY

Megan **Abbott**, *Aurora* (USA)
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