1st 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 16TH

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 AMPAkines 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 17TH

## RESEARCH MEETING: STXB1 IN THE CLINIC

### SESSION III
**The clinical spectrum and standard of care in STXB1-related disorders**
Chairs: A. Garcia Cazorla & M. Milh

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<thead>
<tr>
<th>Time</th>
<th>Title</th>
<th>Speaker(s)</th>
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<tbody>
<tr>
<td>9.00</td>
<td>Multilevel phenotyping of STXB1 patients: the N=You approach</td>
<td>H. Bruining</td>
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<tr>
<td>9.20</td>
<td>Sensory and behavioral profiles in people with STXB1-RD</td>
<td>A. Fetta</td>
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<td>9.40</td>
<td>STXB1 ENGAGE Study: Capturing Patient and Family Lived Experiences</td>
<td>J. Gofshteyn</td>
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<td>10.00</td>
<td>Childrens’ Colorado model of a Neurogenetic Multidisciplinary Clinic</td>
<td>S. Demarest</td>
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<td>10.20</td>
<td>Launch of the Europen STXB1 registry</td>
<td>F. Furia, G. Balagura, H. Stamberger, E. Gardella</td>
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<td>10.40</td>
<td>Coffee Break</td>
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<tr>
<td>11.00</td>
<td>Role of Anti-Seizure Medications in STXB1-RD – Relevance and help from clinical Networking</td>
<td>S. Syrbe</td>
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<td>11.20</td>
<td>The role of 4-Phenyl-Butyrate in STXB1-RD: clinical trial updates</td>
<td>Z. Grinspan</td>
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<td>11.40</td>
<td>Standardized assessments in physical and occupational therapy in STXB1-RD</td>
<td>S. Pierce, K. V. Cunningham</td>
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<td>12.00</td>
<td>Cortical Visual Impairment in STXB1-RD</td>
<td>M. Abbott</td>
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<td>12.20</td>
<td>The emerging scope of neuropsychological abilities in STXB1-RD</td>
<td>A. Miele</td>
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<td>12.40</td>
<td>Lunch</td>
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### SESSION IV
**The future therapeutic landscape in STXB1-related disorders: what to treat, how to measure?**
Chairs: P. Striano & S. Weckhuysen

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<th>Time</th>
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<tr>
<td>14.00</td>
<td>Introduction to the session: targeted therapies for STXB1-RD</td>
<td>J. Goss</td>
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<td>14.20</td>
<td>Clinical trials for rare disorders: challenges and novel strategies</td>
<td>S. Auvin</td>
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<td>14.40</td>
<td>EEG in STXB1-RD: not only epilepsy</td>
<td>E. Gardella</td>
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<td>15.20</td>
<td>Characterizing gait deviations in DEE and potential impact for rehabilitation</td>
<td>A. Hallemans</td>
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<td>15.40</td>
<td>Gut-brain axis in epilepsy and autism</td>
<td>E. Russo</td>
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<td>16.00</td>
<td>Coffee Break</td>
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<td>16.20</td>
<td>Prospective, Multi-Site Natural History Study of STXB1-RD</td>
<td>S. Ruggiero</td>
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<td>16.40</td>
<td>ESCO natural history study</td>
<td>G. Balagura, M. Verhage</td>
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<td>17.00</td>
<td>Anglener Biomarker Outcome Consortium</td>
<td>J. Panagoulis</td>
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<td>17.20</td>
<td>The Loulou Foundation CDKL5 pre-competitive consortium</td>
<td>X. Liogier</td>
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<td>17.40</td>
<td>Virtual clinical trials and seizure prediction in STXB1-RD</td>
<td>J. Xian</td>
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<td>18.00</td>
<td>Roundtable of clinical researchers and industry</td>
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<td>19.00</td>
<td>Closing remarks</td>
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DAY 3 | THURSDAY 18TH

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)
Kristin Cunningham, Philadelphia (USA)
Scott Demarest, Aurora (USA)
Anna Fetta, Bologna (IT)
Francesca Furia, Odense (DK)
Angelas García Cazorla, Barcelona (E)
Elena Gardella, Odense (DK)
Jacqueline Gofshteyn, Philadelphia (USA)
James Goss, Holly Springs (USA)
Zachary Grinspan, New York (USA)
Noah Guiberson, New York (USA)
Anne Hallemans, 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 Panagoulia, Austin (USA)
Jean-Francois Perrier, Copenhagen (DK)
Samuel Pierce, Philadelphia (USA)
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)
Hanna Stamberger, Antwerp (B)
Pasquale Striano, Genova (IT)
Steffen Syrbe, Heidelberg (DEU)
Ruud Toonen, Amsterdam (NL)
Matthijs Verhage, Amsterdam (NL)
Sarah Weckhuysen, Antwerp (B)
Julie Xian, Philadelphia (USA)
Mingshan Xue, Houston (USA)
Federico Zara, Genova (IT)

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