

**PMSDDS 2025**



# CureSHANK 2<sup>nd</sup> Annual Phelan-McDermid Syndrome Drug Development Symposium

Parc de Recerca Biomèdica  
**Barcelona** / June 26-27



Organized by



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PMSDDS 2025

# CureSHANK 2<sup>nd</sup> Annual Phelan-McDermid Syndrome Drug Development Symposium

Thursday June 26, 2025



12:00 – 13:00 h **Registration and light lunch**

13:00 – 13:15 h **Welcoming remarks**

**Geraldine Bliss, MS**

Founder and President – CureSHANK

**Norma Alhambra**

President of the Spanish Phelan-McDermid Syndrome Association

13:15 – 13:30 h **Family stories**

**Sandra Robert**

PMS parent

**Kendra Hegedus, RN**

PMS parent

13:30 – 15:00 h **Introduction to the Phelan-McDermid Syndrome**

MODERATOR:

**Joseph Buxbaum, PhD**

Professor of Psychiatry, Neuroscience and Genetics

Director of the Seaver Autism Center for Research and Treatment

Icahn School of Medicine at Mount Sinai, New York, USA

**SPEAKERS**

13:30 – 13:50 h **Genetics & Epidemiology of PMS**

**Catalina Betancur, MD, PhD**

Director of Research

INSERM, CNRS, Sorbonne University, Paris, France

Chair, Scientific Advisory Board,

Spanish Phelan-McDermid Syndrome Association

13:50 – 14:10 h **Overview of Clinical Features of PMS  
& Natural History Summary**

**Antonio M. Persico, MD**

University of Modena and Reggio Emilia, Italy

## SCIENTIFIC PROGRAM

14:10 – 14:30 h    **Consensus Guidelines, EU Community  
Organization & Trial Readiness**  
**Michael Schön, MD**  
Neuroscientist – Institute for Anatomy and Cell Biology,  
Ulm University, Ulm, Germany  
Chair of the European PMS Research Consortium

14:30 – 14:50 h    **CureSHANK Resources**  
**Ana Mingorance, PhD**  
Chief of Translational Science – CureSHANK

**PANEL**  
14:50 – 15:00 h    **Q&A and Discussion**  
**Catalina Betancur, MD, PhD; Antonio Persico, MD,  
Michael Schön, MD; Ana Mingorance, PhD**

15:00 – 15:30 h    **Afternoon coffee break**

15:30 – 17:30 h    **Clinical outcome measures and biomarkers**

MODERATOR:

**Eva Loth, PhD**  
King's College London, London, England

**SPEAKERS**  
15:30 – 15:50 h    **DSC Natural History Study Outcome Measures**  
**Alex Kolevzon, MD**  
Professor of Child and Adolescent Psychiatry  
Clinical Director of the Seaver Autism Center  
for Research and Treatment  
Icahn School of Medicine at Mount Sinai, New York, USA

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SCIENTIFIC PROGRAM

15:50 – 16:00 h    **PMS-Specific Measure in Development**  
**Alex Kolevzon, MD**

16:00 – 16:20 h    **Neuren Disease-Anchored CGI**  
**Elizabeth Berry-Kravis, MD, PhD**  
Professor, Departments of Pediatrics,  
Neurological Sciences, Anatomy and Cell Biology  
Director, RUSH Pediatric Neurosciences F.A.S.T.  
Center for Translational Research,  
Rush University Medical Center, Chicago, USA

16:20 – 16:40 h    **Fluid Biomarker**  
**Tobias Böckers, MD, PhD**  
Neuroscientist – Institute for Anatomy and Cell Biology, Ulm University  
Ulm, Germany

16:40 – 17:00 h    **Industry Perspectives on Biomarkers**  
**Ayan Ghoshal, PhD**  
Special Adviser to CureSHANK, Cambridge, Massachusetts, USA

17:00 – 17:30 h    **PANEL**  
**Q&A and Discussion**  
**Alex Kolevzon, MD; Liz Berry-Kravis, MD, PhD; Tobias Böckers, MD, PhD; Ayan Ghoshal, PhD**

17:30 – 19:30 h    **Poster reception**

Friday June 27, 2025

08:55 – 09:00 h **Family story**

**Tom Giles**  
PMS parent

09:00 – 10:30 h **Basic & translational research**

MODERATOR:

**Thomas Bourgeron, PhD**  
Institut Pasteur, Paris, France

**SPEAKERS**

09:00 – 09:20 h **Overview of SHANK3**

**Tobias Böckers, MD, PhD**  
Neuroscientist – Institute for Anatomy and Cell Biology, Ulm University  
Ulm, Germany

09:20 – 09:40 h **Development & Validation of Translatable  
Preclinical Assays**

**Joseph Buxbaum, PhD**  
Professor of Psychiatry, Neuroscience and Genetics  
Director of the Seaver Autism Center for Research and Treatment  
Icahn School of Medicine at Mount Sinai, New York, USA

09:40 – 10:00 h **Electrophysiological Biomarkers**

**April Levin, MD**  
Boston Children's Hospital, Boston, USA

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## PANEL

10:00 – 10:30 h

### Discussion

**Dan Gallo, PhD; Morgan Sheng, PhD; Nuno Ribeiro Palha, PhD;  
Tobias Böckers, MD, PhD; Joseph Buxbaum, PhD; April Levin, MD**

#### **Dan Gallo, PhD**

Clinical development and medical affairs, Jaguar Gene Therapy,  
Chicago, USA

#### **Morgan Sheng, PhD**

The Broad Institute of MIT and Harvard,  
Stanley Center for Psychiatric Research, USA

#### **Nuno Ribeiro Palha, PhD**

Research Project Leader, Neurology R&D, Servier, France

10:30 – 10:50 h

### Coffee break

10:50 – 12:40 h

## Noncommercial & Commercial Clinical Trials and Business Considerations

MODERATOR:

#### **Ana Mingorance, PhD**

Chief of Translational Science – CureSHANK

## SPEAKERS

10:50 – 11:00 h

### Lithium Trial

#### **Thomas Bourgeron, PhD**

Institut Pasteur, Paris, France

11:00 – 11:10 h

### Q10 Ubiquinol Trial

#### **Antonio M. Persico, MD**

University of Modena and Reggio Emilia, Italy

## SCIENTIFIC PROGRAM

- 11:10 – 11:20 h      **Intranasal Insulin Trial**  
**Katie Greed**  
PMS parent  
  
**Andre Strydom, MRCPsych, MSc, PhD**  
King's College London, London, England
- 11:20 – 11:35 h      **Jaguar Gene Therapy SHANK3  
Mini-Gene Replacement**  
**Joe Nolan, MBA**  
CEO, Jaguar Gene Therapy, Chicago, USA
- 11:35 – 11:50 h      **Neuren NNZ-2591 for PMS**  
**Elizabeth Berry-Kravis, MD, PhD**  
Professor, Departments of Pediatrics, Neurological Sciences,  
Anatomy and Cell Biology  
Director, RUSH Pediatric Neurosciences F.A.S.T.  
Center for Translational Research, Rush University Medical Center,  
Chicago, USA
- 11:50 – 12:05 h      **PYC SHANK3 ASO**  
**Janya Grainok, PhD**  
Chief Discovery Officer, PYC Therapeutics, Perth, Australia
- 12:05 – 12:20 h      **Q&A**  
**Questions and Answers**

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## PANEL

12:20 – 12:40 h

### Industry Perspectives Discussion

**Jordi Fàbrega, MBA, MS; Jordi Xaus, PhD; Joe Nolan, MBA**

**Jordi Fàbrega, MBA, MS**

Co-Founder and CEO at Connecta Therapeutics, Barcelona, Spain

**Jordi Xaus, PhD**

Chief Scientific Officer, Oryzon, Barcelona, Spain

**Joe Nolan, MBA**

CEO, Jaguar Gene Therapy, Chicago, USA

12:40 – 13:30 h

## Awards Luncheon

13:30 – 15:10 h

## European trial readiness and trial network

MODERATOR:

**Federico Bolognani, MD, PhD**

Chief Medical Officer and co-Founder, Newleos Therapeutics

## SPEAKERS

13:30 – 13:45 h

### Outcome Measures for Rare Diseases

**Silvia Zaragoza Domingo, PhD**

Neuropsychological Research Organization S.L. (Neuropsychro).  
Barcelona, Spain

13:45 – 14:00 h

### EU Registry

**Thomas Bourgeron, PhD**

Institut Pasteur, Paris, France



## SCIENTIFIC PROGRAM

14:00 – 14:15 h     **AIMS-2-TRIALS EAGER Registry**

**Ciara Molloy, PhD**

Senior Research Fellow, Trinity College, Dublin, Ireland  
Research Associate, King's College London, London, UK

14:15 – 14:30 h     **Connect4Children Pan-EU Clinical Trials Network**

**Ricardo Fernandes, MD, PhD**

Chief Medical Officer, conect4children-Stichting, Utrecht, Netherlands

### GROUP DISCUSSION

14:30 – 15:00 h     **Clinical Trials Readiness & Trial Network Infrastructure**

All symposium participants

15:00 – 15:10 h     **Summary of Group Discussion**

**Eric Hoffman, PhD**

CureSHANK Advisor

15:10 – 15:15 h     **Symposium concluding remarks and close**

**Geraldine Bliss, MS**

Founder and President – CureSHANK

**Norma Alhambra**

President of the Spanish Phelan-McDermid Syndrome Association