

**PMSDDS 2025**

**CureSHANK 2nd Annual Phelan-McDermid Syndrome Drug Development Symposium**

**2º Simposio de CureSHANK sobre Desarrollo de Fármacos para el Síndrome de Phelan-McDermid**

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



**Information and online registration at:**  
<https://www.phelanmcdermid-syndrome-global-congress-2025.com/home-symposium-en>

## **PRELIMINARY PROGRAM**

*Please note that the following program is under construction and may be subject to some changes*

### **Thursday June 26, 2025**

**12:00 – 13:00**      **Registration**

**13:00 – 13:15**      **Welcoming remarks**

**Geraline Bliss**

Founder and President – CureSHANK

**Norma Alhambra**

President of the Spanish Phelan-McDermid Syndrome Association

**13:00 – 13:15**      **Family stories**

**Sandra Robert**

PMS parent

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

*Genetics, prevalence, overview of clinical features and natural history, consensus guidelines, industry panel discussion about rare disease drug development*

**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:**

**Catalina Betancur, MD, PhD**

Director of Research  
INSERM, CNRS, Sorbonne University, Paris, France  
Chair, Scientific Advisory Board, Spanish Phelan-McDermid Syndrome Association

**Antonio M. Persico, MD**

University of Modena and Reggio Emilia, Italy

**Michael Schön, MD**

Neuroscientist - Institute for Anatomy and Cell Biology, Ulm University  
Chair of the European PMS Research Consortium

**Ana Mingorance, PhD**

Chief of Translational Science – CureSHANK

**15:00 – 15:30 h**

*Afternoon coffee break*

**15:30 – 17:30 h**

**Clinical outcome measures and biomarkers**

*Clinical outcome measures & biomarkers - natural history/outcome measures, disease-anchored CGI, fluid biomarkers, other biomarker development, panel discussion on COAs and biomarkers.*

**Moderator:**

**Eva Loth, PhD**

King's College London, England

**Speakers:**

**Alex Klevzon, 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

**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

**Tobias Boeckers**

Neuroscientist – Institute for Anatomy and Cell Biology, Ulm University

**17:30 – 19:30 h**

**Poster reception**

## Friday June 27, 2025

09:00 – 10:30 h

### Basic & translational research

*Overview, preclinical assays, electrophysiological biomarkers, panel discussion*

#### Moderator:

**Thomas Bourgeron, PhD**

Institut Pasteur, Paris, France

#### Speakers:

**Tobias Boeckers**

Neuroscientist – Institute for Anatomy and Cell Biology, Ulm University

**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

**April Levin, MD**

Boston Children's Hospital, Boston, US

**Dan Gallo, PhD**

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

**Morgan Sheng**

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

**Nuno Ribeiro**

Research Institute Servier, France

10:30 – 11:00 h

*Coffee break*

11:00 – 12:30 h

### Pipeline

*Noncommercial and commercial clinical trials, regulatory considerations panel*

#### Moderator:

**Ana Mingorance, PhD**

Chief of Translational Science - CureSHANK

#### Speakers:

**Antonio M. Persico, MD**

University of Modena and Reggio Emilia, Italy

**Katie Greed**

PMS Parent

**Andre Strydom, MRCPsych, MSc, PhD**

Kings College, London, UK

**Joe Nolan, MBA**

CEO, Jaguar Gene Therapy, Chicago, US

**Janya Grainok, MSc**

Vice President of Discovery, PYC Therapeutics, Perth, Australia

**12:30 – 13:30 h**

***Awards Luncheon***

**13:30 – 15:10 h**

**European trial readiness and trial network**

***Validation of outcome measures, infrastructure and operational considerations, EU registry, panel discussion about infrastructure models, group discussion or breakout groups***

**Confirmed speakers:**

**Thomas Bourgeron, PhD**

Institut Pasteur, Paris, France

**Silvia Zaragoza, PhD**

Independent consultant, Barcelona, Spain

**15:10 – 15:15 h**

**Symposium concluding remarks and close**

**Geraline Bliss**

Founder and President – CureSHANK

**Norma Alhambra**

President of the Spanish Phelan-McDermid Syndrome Association