

II European Conference Phelan – McDermid Syndrome

June 9, 10 and 11 2023
Monteprincipe campus of CEU San Pablo University
Madrid



Scientific program

Friday June 9th 2023

08:45 – 09:30 h **Registration open**

09:30 – 09:45 h **Opening**

Pablo Lapunzina, MD, PhD

Head of Research Group, INGEMM, La Paz University Hospital Scientific Director, CIBERER, ISCIII, Madrid

Enrique López García

Member of the Board of Directors of FEDER (Spanish Federation of Rare Diseases)

09:45 – 10:00 h **Presentation of the Spanish Phelan-McDermid Syndrome Association**

Norma Alhambra

President of the Spanish Phelan-McDermid Syndrome Association

10:00 – 10:20 h **Testimonials of affected families**

10:20 – 10:55 h **Identification of genotype-phenotype associations in Phelan-McDermid syndrome using family-sourced data from an international registry**

Catalina Betancur, MD, PhD

Director of Research at INSERM, CNRS, Sorbonne University, Paris, France

10:55 – 11:30 h **Clinical and molecular characterization of a large cohort of individuals with Phelan-McDermid syndrome from Spain**

Julián Nevado Blanco, PhD

Head of the Genomics Laboratory
Instituto de Genética Médica y Molecular (INGEMM) La Paz University Hospital, Madrid

11:30 – 12:00 h **Coffee break**

12:00 – 12:40 h **Sensory reactivity in Phelan-McDermid syndrome**

Teresa Tavassoli, PhD

Associate Professor, School of Psychology and Clinical Language Sciences University of Reading, Reading, UK

12:40 – 13:20 h **Preclinical models for drug discovery in Phelan-McDermid syndrome**

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

13:20 – 14:00 h **Inhibition of the cannabinoid CB1 receptor rescues deficits in a mouse model of Phelan-McDermid syndrome**

Flavio Tomasi

PhD student, Sorbonne Université, INSERM, CNRS, Paris, France

Pier Vincenzo Piazza, MD, PhD

Chief executive officer, Aelis Farma, Bordeaux, France

14:00 – 15:00 h **Group photo and lunch**

15:00 – 15:40 h **Developing new treatments and biomarkers in Phelan-McDermid syndrome**

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

15:40 – 16:20 h **A metabolic-based formula improves neurodevelopment in several neurogenetic conditions including Phelan-McDermid syndrome**

Ángeles García Cazorla, MD, PhD

Head of the Neurometabolic Unit and Director of Research in Neurology Hospital Sant Joan de Déu, Barcelona

16:20 – 16:55 h **Unsupervised characterization of Phelan-McDermid syndrome patients: Towards a better-defined clinical trial**

Jordi Xaus, PhD

Chief Scientific Officer, Oryzon, Barcelona

16:55 – 17:30 h **What CureSHANK is doing to bridge gaps in drug development for Phelan-McDermid syndrome**

Paulina Rychenkova, PhD

Co-founder and Secretary, CureSHANK, USA

17:30 – 19:00 h **Poster session with beer and tapas**

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Saturday June 10th 2023

09:15 – 09:40 h **Registration open**

09:40 – 09:45 h **Welcome speech**

Norma Alhambra

President of the Spanish Phelan-McDermid Syndrome Association

09:45 – 10:45 h **Current research in Phelan-McDermid syndrome & scientific day highlights**

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

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

Catalina Betancur, MD, PhD

Director of Research at INSERM, CNRS, Sorbonne University, Paris, France

10:45 – 11:30 h **Neuropsychiatric symptoms and loss of skills in Phelan-McDermid syndrome**

Alex Kolevzon, MD

11:30 – 12:00 h **Coffee break**

12:00 – 12:40 h **Sleep issues in Phelan-McDermid syndrome**

Antonio Persico, MD

Professor of Child and Adolescent Psychiatry University of Modena and Reggio Emilia, Modena, Italy

12:40 – 13:20 h **Genetic defects in Phelan-McDermid syndrome**

Catalina Betancur, MD, PhD

13:20 – 14:00 h **Neurofibromatosis and ring chromosome 22**

Claire Forde, MD

Consultant Clinical Geneticist, Manchester Centre for Genomic Medicine, Manchester University, Manchester, UK

14:00 – 15:30 h **Group photo and lunch**

Concurrent activity: Understanding your genetic reports. One-on-one meeting with a geneticist who will explain your genetic reports to you. By appointment only

Concurrent activity: Poster session

15:30 – 16:40 h **Presentation of the European medical guidelines on Phelan-McDermid syndrome prepared by the European Phelan-McDermid Syndrome Consortium**

Conny van Ravenswaaij-Arts, MD, PhD

Professor in Dysmorphology, Department of Genetics University of Groningen, Groningen, the Netherlands

Michael Schön, MD, PhD

Research Assistant, Institute for Anatomy and Cell Biology Ulm University, Ulm, Germany

Alison Turner

Parent representative
Phelan-McDermid Syndrome Foundation UK

16:40 – 17:25 h **Consensus guidelines on the management of Phelan-McDermid syndrome: results from an international working group**

Alex Kolevzon, MD

17:25 – 18:00 h **Panel discussion and questions**

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Sunday June 11th 2023

- 10:00 – 10:50 h **Epilepsy in Phelan-McDermid syndrome**
Sarah Jesse, MD
Department of Neurology, Ulm University, Ulm, Germany
- 10:50 – 11:25 h **Sensory processing traits: evaluation and evolution in Phelan-McDermid syndrome**
Sergio Serrada Tejada, PhD
Department of Physical Therapy, Occupational Therapy, Physical Medicine and Rehabilitation, King Juan Carlos University, Alcorcón, Spain
- 11:25 – 11:55 h **Coffee break**
- 11:55 – 12:30 h **Autistic symptoms in Phelan-McDermid syndrome: comparison with idiopathic autism and longitudinal study**
Mónica Burdeus
Psychologist, Department of Psychiatry
Hospital General Universitario Gregorio Marañón, Madrid
- 12:30 – 13:05 h **Understanding and accompanying siblings of individuals with Phelan-McDermid syndrome in their growth and relationships**
Stefano Cainelli, PhD
Laboratory of Observation and Diagnosis and Training
University of Trento, Trento, Italy
- 13:05 – 13:25 h **Intestinal microbiota in patients with Phelan-McDermid syndrome and its modulation through the use of a probiotic**
Juan Miguel Rodríguez Gómez, PhD
Nutrition and Food Science Department, Complutense University, Madrid
- 13:25 – 13:55 h **Panel discussion and questions**
- 13:55 – 14:00 h **Closure of the conference**
Norma Alhambra
President of the Spanish Phelan-McDermid Syndrome Association
- 14:00 – 15:30 h **Lunch**