

PHELAN-MCDERMID SYNDROME INTERNATIONAL CONFERENCE EUROPE

Madrid 21-23 September 2018

Ortiz-Vázquez Auditorium. La Paz University Hospital



Information and online registration in:

<http://eventos.aymon.es/conferencia-internacional-phelanmcdermid/>

<http://www.22q13.org.es/conferencia-2018/>

SCIENTIFIC PROGRAM

FRIDAY, SEPTEMBER 21

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|-----------------|--|
| 08:45 – 09:30 h | Registration open – Registration desk |
| 09:30 – 09:45 h | Opening |
| 09:45 - 10:00 h | Presentation of the Spanish Phelan-McDermid Syndrome Association
<i>Norma Alhambra</i> |
| 10:00 - 10:15 h | Testimonials of affected families □ |
| 10:15 - 10:45 h | Overview of microdeletion syndromes
<i>Pablo Lapunzina, MD, PhD</i> |
| 10:45 - 11:30 h | Clinical and genetic spectrum of Phelan-McDermid syndrome
<i>Catalina Betancur, MD, PhD</i> |
| 11:30 - 11:55 h | Coffee break |
| 11:55 - 12:40 h | Cell and animal models as a means to develop novel treatments in Phelan-McDermid syndrome
<i>Joseph D. Buxbaum, PhD</i> |
| 12:40 - 13:25 h | From model systems to clinical trials in children with Phelan-McDermid syndrome
<i>Alexander Kolevzon, MD</i> |

- 13:25 - 14:10 h **Neuropsychiatric phenotype and psychopharmacology in patients with Phelan-McDermid syndrome**
Willem M. A. Verhoeven, MD
- 14:10 - 15:15 h **Group photo and lunch**
- 15:15 - 16:00 h **Characterising the neurocognitive profile of people with Phelan McDermid Syndrome and the relationship to individual differences in clinical features**
Eva Loth, PhD
- 16:00 - 16:45 h **New approaches to the study of genotype/phenotype correlations in a cohort of 138 mainly Spanish individuals with PMS**
Julián Nevado, PhD
- 16:45 - 17:30 h **Developmental trajectories, autistic features and psychiatric comorbidities in 50 Spanish patients with PMS**
Mónica Burdeos
Mara Parellada, MD, PhD
- 17:30 - 19:00 h **Poster session with beer and tapas**

SATURDAY, SEPTEMBER 22

- 09:15 - 09:45 h **Registration open – Registration desk**
- 09:45 - 09:55 h **Wellcome speech**
Norma Alhambra
President of the Spanish PMS Association
- 09:55 - 10:25 h **PMS in the world**
Sue Lomas
President of the PMS Foundation
- 10:25 - 11:05 h **PMS Across the Lifetime**
Alexander Kolevzon, MD
- 11:05 - 11:45 h **Genetics of PMS**
Catalina Betancur, MD, PhD
- 11:45 - 12:10 h **Coffee break**
- 12:10 - 12:50 h **Managing epileptic seizures in PMS**
Antonio Gil-Nagel, MD, PhD
- 12:50 - 13:30 h **Contextual neuropsychological approach in neurodevelopmental disorders**
Jos I. M. Egger, PhD
- 13:30 - 14:10 h **The role of microbiota in autism spectrum disorder**
Guillermo Alvarez Calatayud, MD, PhD

- 14:10 - 15:40 h **Group photo and lunch**
Concurrent activity: Understanding your genetic reports. One-on-one meeting with a geneticist who will explain your genetic report.
By appointment only
- 15:40 - 16:20 h **Diagnostic and therapeutic aspects of pediatric lymphedema**
Juan Carlos López Gutierrez, PhD
- 16:20 - 17:40 h **Current research in Phelan-McDermid syndrome & Scientific meeting day highlights**
Joseph D. Buxbaum, Alex Kolevzon, and Catalina Betancur
- 17:40 - 18:20 h **Gene therapy: challenges and promises**
Lluís Montoliú, PhD

SUNDAY, SEPTEMBER 23

- 10:00 - 10:40 h **Best clinical practices for PMS**
Alexander Kolevzon, MD □
- 10:40 - 11:20 h **Sleep problems in PMS**
Rafael del Río, MD
- 11:20 - 11:45 h **Puberty problems in PMS**
David Fraguas, MD, PhD
- 11:45 - 12:10 h **Coffee break**
- 12:10 - 12:35 h **Long term treatment with IGF-1 in a girl with PMS: a case study**
Mara Parellada, MD, PhD
- 12:35 - 13:45 h **Discussion panel with all speakers, summary and questions**
Moderator: Katy Phelan, PhD
- 13:45 - 14:00 h **Closure of the conference**
Norma Alhambra
- 14:00 - 15:30 h **Lunch**