

**SAVE THE
DATE**

MADRID

May 10-11, 2018



International Symposium on Somatic Mosaicism in Vascular Malformations

This symposium will bring together an international panel of experts that will offer a global and interdisciplinary vision on the etiopathological, clinical and diagnostic aspects of vascular malformations associated with somatic mosaicism.

Venue:

Fundación Ramón Areces

c/ Vitruvio, 5. 28006 Madrid

Free access.

Online registration is kindly requested

<http://www.fundacionareces.es>

Program

May 10

- 09:30 - 09:45 Welcome
- 09:45 - 10:15 **Mosaicism in health and disease.** Lars A. Forsberg. Department of Immunology, Genetics and Pathology, Uppsala University, Uppsala SE-75108, Sweden.
- 10:15 - 10:45 **Overview of somatic mosaicism in vascular malformations.** Víctor Martínez-Glez. Institute of Medical and Molecular Genetics (INGEMM), IdiPAZ, Hospital Universitario La Paz, Madrid, Spain; CIBERER, Instituto de Salud Carlos III, Madrid, Spain.
- 10:45 - 11:15 **Clinical classification: controversies and recent advances.** Juan Carlos Lopez-Gutierrez. Vascular Anomalies Unit, Hospital Universitario La Paz. Madrid, Spain.
- 11:15 - 11:45 **A fresh look at capillary malformations: why do we need particular names for different vascular skin lesions?** Rudolf Happle. Institute of Human Genetics, University Hospital Magdeburg, Otto-von-Guericke University, Magdeburg, Germany.
- 11:45 - 12:00 Coffee break
- 12:00 - 12:30 **Histopathological review of vascular malformations associated to somatic mosaicism.** Isabel Colmenero. Paediatric histopathologist. Hospital Niño Jesús. Madrid, Spain.
- 12:30 - 13:00 **Mosaicism in Arteriovenous malformations.** Veronica Kinsler. Paediatric Dermatology, Great Ormond St Hospital for Children, London, U.K.
- 13:00 - 14:00 Roundtable: Chair: Juan Carlos Lopez-Gutierrez
- 14:00 - 16:00 Lunch
- 16:00 - 16:30 **Selected clinical cases.** Marta Feito. Dermatology Service, Hospital Universitario La Paz, Madrid, Spain.
- 16:30 - 17:00 **Combined vascular and non-vascular mosaic anomalies.** Antonio Torrelo. Department of Dermatology, Hospital Infantil del Niño Jesús, Madrid, Spain.
- 17:00 - 17:30 **NGS and phenotyping of mosaic skin development anomalies.** Pierre Vabres. Service de Dermatologie, CHU Dijon Bourgogne, Dijon, France.
- 17:30 - 18:00 **Bioinformatic challenges in the detection of low mosaics by NGS.** Kristina Ibañez. Institute of Medical and Molecular Genetics (INGEMM), IdiPAZ, Hospital Universitario La Paz, Madrid, Spain; CIBERER, Instituto de Salud Carlos III, Madrid, Spain.
- 18:00 - 18:30 Roundtable: Chair: Sixto Garcia Miñaur
Welcome

May 11

- 09:30 - 10:00 **PIK3CA-related overgrowth spectrum (PROS).** Leslie Biesecker. National Human Genome Research Institute, National Institutes of Health, Bethesda, Maryland, USA.
- 10:00 - 10:30 **New PIK3CA-related syndromes.** Lara Rodriguez-Laguna. Institute of Medical and Molecular Genetics (INGEMM), IdiPAZ, Hospital Universitario La Paz, Madrid, Spain; CIBERER, Instituto de Salud Carlos III, Madrid, Spain.
- 10:30 - 11:00 **Advances in animal models and treatment perspectives in PIK3CA-associated lymphatic malformations.** Mike Dellinger. Department of Surgery, University of Texas Southwestern Medical Center, Dallas, TX, USA.
- 11:00 - 11:30 **Modeling somatic mutations in mice.** Matthew Warman. Vascular Biology Program and Department of Surgery, Boston Children's Hospital, Harvard Medical School, Boston, USA.
- 11:30 - 12:00 Coffee break
- 12:00 - 12:30 **Using CRISPR editing tools to counteract somatic mosaicism.** Lluís Montoliu. Department of Molecular and Cellular Biology, Centro Nacional de Biotecnología (CNB-CSIC), Madrid, Spain; CIBERER, Instituto de Salud Carlos III, Madrid, Spain.
- 12:30 - 13:00 **Genetic counselling for mosaic disorders.** Sixto Garcia Miñaur. Institute of Medical and Molecular Genetics (INGEMM), IdiPAZ, Hospital Universitario La Paz, Madrid, Spain; CIBERER, Instituto de Salud Carlos III, Madrid, Spain.
- 13:00 - 13:30 **What should we keep in mind when talking about somatic mosaicism?** Pablo Lapunzina. Institute of Medical and Molecular Genetics (INGEMM), IdiPAZ, Hospital Universitario La Paz, Madrid, Spain; CIBERER, Instituto de Salud Carlos III, Madrid, Spain.
- 13:30 - 14:00 Roundtable: Víctor Martínez-Glez
- 14:00 Closure