



PVRI
Pulmonary Vascular
Research Institute



Date // **4-5 February 2019**

1st International Consortium for Genetic Studies in PAH

Featuring the PVRI Pre-Clinical & Molecular Science Task Force

Hotel // **The Crowne Plaza Hotel · Barcelona**

BARCELONA
2019

Title: **1st International Consortium for Genetic Studies in PAH**

Day 1 4 February

Chairs:

- // 09:00: Nick Morrell & Mandy MacLean
 - // 11:15: Micheala Aldred & Pilar Escribano
 - // 14:00: Wendy Chung & Raymond Benza
 - // 16:15: Ekkehard Grünig & Florent Soubrier
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09:00	Welcome & introduction // Nick Morrell
09:15	Keynote: translational genome annotation with human gene knockouts // Richard Trembath
09:45	Genetics of PAH across the lifespan // Wendy Chung
10:05	Identification of new PAH genes // Stefan Gräf
10:25	PAH genes in the US PAH Biobank // William Nichols
10:45	Coffee
11:15	Gene defects in high altitude pulmonary oedema susceptibles // Ekkehard Grünig et al
11:35	The BMP10 gene widens the landscape of heritable PAH mutations // Melanie Eyries et al
11:50	Genetic analysis in a lung transplant cohort (PHBI) // Micheala Aldred
12:10	Common variant analysis in PAH // Chris Rhodes
12:30	Lunch
14:00	PVDomics update // Anna Hemnes
14:20	Association of common variants with outcomes and phenotypes in PAH // Ankit Desai
14:40	PAH associated to congenital heart disease: something more than a structural defect // Carmen Perez-Olivares et al

14:55	Clinical phenotype and prognosis of PAH patients with TBX4 mutations // Ignacio Hernandez-Gonzalez et al
15:10	Genetic linkage analysis of a large family identifies FIGN as a candidate modulator of reduced penetrance in heritable PAH // Pau Puigdevall et al
15:25	PAH gene panel: a new diagnostic tool for patients with PAH // Christina Eichstaedt et al
15:40	Coffee
16:15	Investigating the contribution of SOX17 mutations to the pathogenesis of PAH // Joy Shih et al.
16:30	Transcriptomic profiles associate with PAH severity, outcomes and endothelial activation // Casey Romanoski et al.
16:45	Germline and acquired dysregulation of TET2 and DNMT3A in PAH // François Potus et al
17:00	Summary and close of day 1 // Nick Morrell

Day 2

5 February

Chairs:
 // 09:00: Richard Trembath & Martin Wilkins
 // 11:10: Stephen Archer & Werner Seeger
 // 14:00: Roham Zamanian & Eric Austin

09:00	Keynote: making sense of non-coding genome variation – lessons from diabetes // Jorge Ferrer
09:30	Phenotypic diversity among paediatric PAH with TBX4 mutations // Eric Austin
09:50	Genetics and mechanisms of PVOD // David Montani
10:10	Analysis of the BMPR2 promoter // Rizwan Hamid
10:30	RNA-seq of peripheral blood in PAH // Dennis Wang & Allan Lawrie
10:50	Coffee

11:10	Integrating omics for intermediate phenotypes // Dr Lars Harbaum
11:30	Gene editing human pluripotent stem cells to understand the contribution of genetic and environmental factors in PAH // Amer Rana
11:50	Genetic admixture and clinical profiles in minority populations with PAH // Jason Karnes et al
12:05	Customized massive parallel sequencing panel for diagnosis of pulmonary arterial hypertension // Jair Tenorio et al
12:20	Lunch & posters
14:00	Poster summaries (1 slide, 2 minutes each!) Genetic diagnostics in patients with CTEPH // Christina Eichstadt et al Different forms of PH in a family with clinical and genetic evidence for HHT type 2 // Alessandro Greco et al HHT related mutations in a Spanish population of patients with PAH // Agueda Arunetxeta et al Identification of novel genetic markers associated to PAH in connective tissue disease // Carmen Perez-Olivares Genetic analysis in PAH Sicilian patients: preliminary results // Italia Loddo et al Novel bone morphogenic protein receptor 2 mutation in a patient with hereditary pulmonary arterial hypertension // Shine Kumar et al Detection of latent pulmonary hypertension in genetically susceptible individuals (DOLPHIN-GENESIS) // Samara Jansen et al Patient affected with both pulmonary arterial hypertension and hereditary hemorrhagic telangiectasia carry a novel ENG intronic variant affecting gene transcription // Carla Olivieri et al
14:30	Aims and objectives of the PAH-ICON collaboration Discussion and consideration of specific projects
17:00	Close



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