



PVRI
Pulmonary Vascular
Research Institute



Date // **2-3 February 2020**

The 2nd International Consortium Meeting for Genetic Studies in Pulmonary Arterial Hypertension (PAH)

Hotel // **The Swissotel · Lima · Peru**

This meeting has been kindly sponsored by
the Pulmonary Vascular Research Institute

LIMA
2020

Title: **The 2nd International Consortium Meeting for Genetic Studies in Pulmonary Arterial Hypertension (PAH)**

Day 1 2 February

14:00 **Welcome and introduction**
// Nick Morrell UNIVERSITY OF CAMBRIDGE, UK

14:15 **Proteome/Metabolomics Task Force**
// Martin Wilkins IMPERIAL COLLEGE LONDON, UK

14:55 **Abstract: Whole exome sequencing analysis of idiopathic and heritable pulmonary arterial hypertension**
// Jair Tenorio 12 OCTUBRE HOSPITAL, SPAIN
(On behalf of the Hospital 12 de Octubre)

15:10 **Abstract: Genetic findings in patients with different forms of pulmonary hypertension**
// Christina Eichstaedt HEIDELBERG UNIVERSITY HOSPITAL, GERMANY

15:25 **Break**

15:50 **Clinical Phenotypes Task Force**
// Roham Zamanian STANFORD UNIVERSITY, USA

16:30 **Abstract: Heritable veno-occlusive pulmonary disease**
// Jair Tenorio INGEMM-HULP, SPAIN
(On behalf of the Hospital 12 de Octubre)

16:45 **Abstract: The combination of metabolomic and miRNA diagnostic scores to identify pulmonary hypertension and sub-classify pulmonary arterial hypertension**
// Chris Rhodes IMPERIAL COLLEGE LONDON, UK

17:00 **Mitochondrial Genetics Task Force**
// Micheala Aldred INDIANA UNIVERSITY, USA

END OF DAY 1

Day 2 **3 February**

09:00 **RNA sequencing Task Force**
// Chris Rhodes IMPERIAL COLLEGE LONDON, UK

09:40 **Abstract: Rare and de novo variants identified in 4,043 PAH cases from an international consortium implicate new PAH candidate genes**
// Wendy Chung & Yufeng Shen COLUMBIA UNIVERSITY, USA

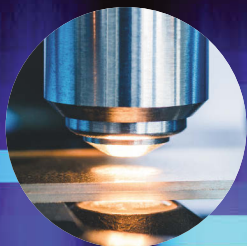
09:55 **Abstract: The role of sorting nexin 29 (SNX29): a novel biomarker for vasoresponsive pulmonary arterial hypertension (PAH)**
// Jason Karnes INDIANA UNIVERSITY, USA

10:10 **Epigenetics Task Force**
// Stephen Archer & François Potus QUEEN'S UNIVERSITY, CANADA

10:50 **Break**

- 11:20 **Abstract: Familial pulmonary arterial hypertension by KDR heterozygous loss of function**
// Florent Soubrier HÔPITAUX DE PARIS, FRANCE
-
- 11:35 **TBX4 Task Force**
// Eric Austin VANDERBILT UNIVERSITY, USA
-
- 12:15 **Abstract: Rare variant association study of multiple pulmonary hypertension phenotypes using a Bayesian statistics framework**
// Emilia Swietlik UNIVERSITY OF CAMBRIDGE, UK
-
- 12:30 **EIFAK4 Task Force**
// Greg Elliott INTERMOUNTAIN HEALTHCARE
// David Montani HÔPITAUX DE PARIS, FRANCE
-
- 13:00 **Lunch**
-
- 14:00 **Healthy Mutation Carriers Task Force**
// Harm Jan Bogaard AMSTERDAM UNIVERSITY MEDICAL CENTER, THE NETHERLANDS
-
- 14:40 **ClinGen Task Force**
// Wendy Chung COLUMBIA UNIVERSITY, USA
-
- 15:20 **Abstract: Mitochondrial haplogroup analysis in PAH**
// Micheala Aldred INDIANA UNIVERSITY, USA
-
- 15:35 **Vasoresponders Task Force**
// Ankit Desai INDIANA UNIVERSITY, USA
-
- 16:15 **Scleroderma Task Force**
// Ankit Desai INDIANA UNIVERSITY, USA
-

END OF DAY 2



The 2nd International Consortium Meeting for Genetic Studies in Pulmonary Arterial Hypertension (PAH)

LIMA 2020



PVRI

Pulmonary Vascular
Research Institute

www.pvrinstitute.org

The PVRI is a Registered Charity in the UK, No: 1127115.

Disclaimer: This programme and its contents are correct at the time of printing.