



PVRI Genetics & Molecular Report 2019

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Over 140 delegates attended the inaugural PAH-ICON Symposium in Barcelona.

1st Symposium for the International Consortium of Genetic Studies in PH.

The meeting consisted of two days of presentations by invited speakers in the field of genetics and genomics of PAH.

In addition, the meeting included oral and poster abstract presentations by investigators from around the world. The audience learned about recent advances in PAH genetics, including the first announcement of three new genes underlying PAH.

On the afternoon of the second day, the Consortium discussed aims and objectives for the coming year that would be best tackled by international collaboration and networking in this rapidly evolving field. Several Task Forces were established, whereby international collaborators will work together around priority areas. It was agreed that the group will meet

annually to share results and to enable further networking.

Topics presented included:

- Genetics of PAH across the lifespan.
- Common variant analysis in PAH.
- RNA-sequencing of peripheral blood in PAH.

Examples of Task Forces

- Genetics of paediatric PAH.
- ClinGen PAH working group to curate causal PAH mutations.
- Functional genomics Task Force.
- Establish international registry of PVOD cases with EIF2AK4 mutations.
- Follow up of healthy BMPR2 mutation carriers.
- Sequencing of BMPR2 negative PAH families.
- Twin studies in PAH.

It is clear that only international collaborations, which are capable of generating thousands of samples from patients with idiopathic and heritable PAH, will have the statistical power to discover the complete genetic architecture of PAH and to address the major questions regarding the role of genetic variation in disease penetrance, phenotype and the clinical course of disease.

The consortium aims to address these questions and continues to meet regularly by video conferencing to address these areas.

There was a real sense in the meeting that we can better address the major outstanding questions in our field by working together collaboratively for the benefit of patients, scientific discovery and the search for new treatments.

