Special Issue

Genetics and Genomics of Pulmonary Arterial Hypertension

Message from the Guest Editors

Pulmonary arterial hypertension (PAH) is a severe and progressive vascular disorder, which typically manifests at around 30-40 years of age and is often fatal. Pathogenic mutation of the bone morphogenetic protein receptor type 2 (BMPR2) gene is the major PAH risk factor, with over 480 distinct variants reported to date. However, PAH demonstrates substantial locus heterogeneity and missing heritability, particularly in sporadic disease. With the advent of next-generation sequencing, there has been an explosion of new findings in PAH, expanding the genetic architecture of idiopathic disease and highlighting numerous additional risk factors and novel pathways for exploration. In this Special Issue, we aim to provide an overview of the advancements in PAH genetics and genomics over the past decade, and to examine the emerging interplay between genotype and phenotypic outcomes. We also seek to discuss the role of epigenetics and common variation in the pathogenesis of PAH.

Guest Editors

Dr. Laura Southgate

Molecular and Clinical Sciences Research Institute, St George's University of London, London, UK

Dr. Rajiv Machado

Molecular and Clinical Sciences Research Institute, St George's University of London, London, UK

Deadline for manuscript submissions

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Genes
Editorial Office
MDPI, Grosspeteranlage 5
4052 Basel, Switzerland
Tel: +41 61 683 77 34
genes@mdpi.com

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Genes is central to our understanding of biology, and modern advances such as genomics and genome editing have maintained genetics as a vibrant, diverse and fast-moving field. There is a need for good quality, open access journals in this area, and the Genes team aims to provide expert manuscript handling, serious peer review, and rapid publication across the whole discipline of genetics. Starting in 2010, the journal is now well established and recognised. Why not consider Genes for your next genetics paper?

Editor-in-Chief

Prof. Dr. Selvarangan Ponnazhagan

Department of Pathology, The University of Alabama at Birmingham, 1825 University Blvd, SHEL 814, Birmingham, AL 35294-2182, USA

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