



genes



an Open Access Journal by MDPI

Genetics of Congenital Heart Diseases

Guest Editors:

Dr. Jiuann-Huey Ivy Lin

1. Department of Critical Care Medicine, University of Pittsburgh, Pittsburgh, PA 15224, USA

2. Department of Developmental Biology, University of Pittsburgh, Pittsburgh, PA 15224, USA

Dr. Mousumi Moulik

Divisions of Cardiology, and Medical Genetics & Genomic Medicine, Department of Pediatrics, University of Pittsburgh, Pittsburgh, PA 15260, USA

Deadline for manuscript submissions:

closed (5 April 2025)

Message from the Guest Editors

Congenital heart diseases (CHDs) are the most common birth defect, affecting about 1% of neonates. A genetic contribution is strongly implicated in the pathogenesis of CHDs. Copy-number variations and chromosomal anomalies/aneuploidy are found in 8 to 25% of CHD patients. The largest genetic study of CHD from the Paediatric Cardiac Genomics Consortium (PCGC) using whole-exome sequencing identified that 8% of individuals are associated with de novo autosomal dominant variation and 2% of cases are attributed to autosomal recessive variation. There is mounting evidence that therapies targeting the treatment of adult heart failure have been ineffective in the treatment of heart failure in patients with CHD. It implies different pathomechanisms of congestive heart failure in patients with CHD and structurally normal hearts. Therefore, there is an urgent and increasing need to understand the genetic basis of CHDs for precise diagnosis, identifying at-risk patients before clinical symptoms develop, appropriate management, the determination of prognosis, and estimation of the risk of recurrence. This Special Issue is focused on the genetic contribution to congenital heart defects.



mdpi.com/si/136354

Special Issue



genes



an Open Access Journal by MDPI

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

Message from the Editor-in-Chief

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?

Author Benefits

Open Access: free for readers, with article processing charges (APC) paid by authors or their institutions.

High Visibility: indexed within Scopus, SCIE (Web of Science), PubMed, MEDLINE, PMC, Embase, PubAg, and other databases.

Journal Rank: JCR - Q2 (Genetics and Heredity) / CiteScore - Q2 (Genetics (clinical))

Contact Us

Genes Editorial Office
MDPI, Grosspeteranlage 5
4052 Basel, Switzerland

Tel: +41 61 683 77 34
www.mdpi.com

mdpi.com/journal/genes
genes@mdpi.com
[X@Genes_MDPI](https://twitter.com/Genes_MDPI)