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Genetics of Congenital Heart Diseases

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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.



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Special Issue



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Message from the Editor-in-Chief

Genes are central to our understanding of biology, and modern advances such as genomics and genome editing have maintained genetics as a vibrant, diverse and fastmoving 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.

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