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Hypertrophic Cardiomyopathy: Genetics, Pathogenesis, Clinical Manifestations, Diagnosis, and Therapy

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Message from the Guest Editors

Hypertrophic cardiomyopathy (HCM) is the most common inherited cardiovascular disease caused by mutations in sarcomeric proteins. It is characterized by increased ventricular wall thickness and is highly complex due to its heterogeneous clinical presentation, several phenotypes, large number of associated causal mutations, and broad spectrum of complications, such as heart failure and sudden death. There is no curative treatment for HCM, as current therapies are focused on relieving symptoms by pharmacological intervention and not on the cause of HCM. In the last decade, several strategies have been developed to remove genetic defects, including genome editing, exon skipping, allele-specific silencing, spliceosome-mediated RNA trans-splicing, and gene replacement. Most of these technologies have already been tested for efficacy and efficiency in animal- or human-induced pluripotent stem cell models of HCM with promising results. Therefore, the aim of this Special Issue is to highlight the most recent advances in the field of HCM, including diagnosis and clinical management of mixed phenotypes, genetics, and new therapies.

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