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Genetic Basis for Congenital Heart Disease

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Abstract

This article presents a current overview of our understanding of genetic contributions to the development of congenital cardiac disease. Since the year 2007 the publication of a paper on the genetic basis of congenital heart disease, new genomic tools have been widely available, drastically altering our understanding of the aetiology of congenital heart disease. New molecular testing techniques are discussed, as well as their use to congenital cardiac disease, both alone and in combination with other congenital defects or syndromes. The latest research on copy number variations, syndromes, RASopathies, and heterotaxy/ciliopathies is presented. New research findings using congenital heart disease models. This review is expected to provide timely information on the genetic aspects of congenital heart disease to a wide range of health-care professionals, including paediatric cardiologists, paediatricians, adult cardiologists, thoracic surgeons, obstetricians, geneticists, genetic counsellors, and other related clinicians.

Keywords: Congenital defects • Cardiac disease • Cardiologists

Editorial

Hypertension is one of the leading causes of premature morbidity and mortality in the world. In 2013, it was identified as the second most important risk factor for the global burden of disease. Ischemic and haemorrhagic stroke, myocardial infarction, heart failure, chronic renal disease, peripheral vascular disease, cognitive decline, and early death are all linked to hypertension. Moreover, a vast number of observational studies have found a graded independent connection between systolic and diastolic blood pressures and mortality and morbidity. Untreated hypertension can lead to a steady rise in blood pressure, potentially leading to treatment resistance owing to vascular and renal damage. In a population, blood pressure is distributed regularly, and there is no natural cut off point over which hypertension is certainly present and below which it is not. Systolic and/or diastolic blood pressures may be raised in any given person. People under the age of 50 are more likely to have high diastolic pressure. Systolic hypertension becomes more of an issue as people get older, due to the gradual stiffness and loss of compliance of bigger arteries. This review was written to inform clinicians about recent advances in our understanding of genetic factors to the aetiology of congenital heart disease as well as to offer an update on the 2007. The discovery of multiple harmful Copy Number Variants (CNVs) and gene mutations has substantially increased our understanding of the causes of Congenital Heart Disease since the rapid proliferation of these and other testing methods mentioned in this study. The impact of excellent medical and surgical care of congenital HD on individual survival is likely contributing to the rising prevalence of congenital HD among older children and adults to a great extent. Patients with severe forms of congenital HD are increasingly living into their 30s and beyond. According to epidemiological research, a genetic or environmental aetiology can be found in 20% to 30% of congenital heart disease cases.

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