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Advances in Cardiology and Cardiovascular Disorders

Recent research in the area of cardiac genetics and genomics

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Cardiac genetics and genomics are important facts of innovation in current scenario. There are various studies done in this field to identify the abnormalities and treatment of cardiac conditions. The aim is to find out the different researches in the various age groups regarding congenital heart diseases, risk factors, testing and treatment modalities of cardiovascular diseases (CVD).

Schematic Representation of Research in Cardiac Genetics and Genomics

Fetus: Congenital heart disease (CHD) is the most common birth defect. The concept of second heart field, augment the initial simple heart tube, has clarified the development of the heart. Understanding how this is used in morphogenesis and how genes interact in a subtle and more complex way is moving us closer to understand how the normal heart forms and why abnormalities occur.

Infant: Whole-exome sequencing (WES) represents an unbiased screening of the exome, which could help to investigate different pathogenic mechanisms within the genetically heterogeneous Sudden infant death syndrome (SIDS) cohort. Additionally, re-analysis of the datasets provides the basis to identify new candidate genes in SIDS.

Children: Copy number changes in genes such as GATA4 and NODAL can contribute to CHD and CDK13 protein produced by novel gene with causative mutations accounts for newly discovered CHD

Adults: Elevated gene expressions of TRP channels are associated with the pathogenesis of Non-valvular atrial fibrillation (NVAF). COL21A1 and RRAS warrant particular interest since both are involved in blood vessel remodelling, with relevance to hypertension. Mutation in a key molecule ENPEP affects blood pressure. This gene codes for an enzyme that is a key molecule involved in regulating blood pressure through the dilation and constriction of blood vessels.

Old age: Chromosomes 4 and 7 are associated with extreme survival and with reduced risk for cardiovascular disease.

Death: In genotype-positive family members, an intracardiac defibrillator is implanted to reduce the risk of sudden cardiac death.

Treatment: Human induced pluripotent stem cell (hiPSC)-derived cardiomyocytes can serve to enhance our understanding of the development, pathophysiology and potential therapeutic targets for CHD.

Genetic counselling and testing: Genetic counselling strategies for transforming the public opinion regarding the practice of consanguinity and its associated risks. It is highly useful for cardiomyopathies, arrhythmia syndromes such as Long QT and Brugada, and inherited aortopathies including connective tissue disorders such as Marfan, Ehlers-Danlos, and Loeys- Dietz syndromes. With newer agents targeting proprotein convertase subtilisin/kexin type 9 inhibition, it may be necessary to document a genetic diagnosis of familial hypercholesterolemia prior to prescribing.

Genetics and genomics serve several roles in cardiovascular health and disease, including disease prediction, discovery of genetic loci influencing CVD and prevention strategies.