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Mutations of Cardiovascular Diseases

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Introduction

Myocardial infraction to congenital heart disease are all examples of cardiovascular illness, the majority of which are heritable. A great deal of research has gone into figuring out which genes and DNA sequence variants are responsible for this heritability. Cardiovascular disease (CVD) is a major health issue that affects approximately 80 million people in the United States alone. CVD refers to a wide range of conditions, including diseases of the vascular, myocardium, the electrical circuit of the heart, and congenital heart disease. Inherited DNA sequence variants play a role in illness risk for nearly all of these disorders. A major goal of biomedical research over the past century has been to connect genotype with phenotype, that is, to find the exact genes and DNA sequence variants that cause characteristic variation in humans.

Description

This is especially essential because some diseases, like as myocardial infarction (MI), are difficult to simulate in non-human species. A major goal of biomedical research over the past century has been to connect genotype with phenotype, that is, to find the exact genes and DNA sequence variants that cause characteristic variation in humans. What is the primary motivation for pursuing this goal? Natural genetic diversity in humans has the unique ability to reveal causative biologic pathways. This is especially essential because some diseases, like as myocardial infarction (MI), are difficult to simulate in non-human species.

Hypertrophic cardiomyopathy (HCM) was the first hereditary cardiovascular ailment to have a genetic basis, and as a result, it has served as a model for how to investigate a genetic cardiovascular disease. Despite substantial progress in identifying the genetic origins of HCM, current data reveal that a significant proportion of HCM patients have several mutations. More than 400 mutations in at least 13 genes have been discovered in HCM patients since 1990. The two most prevalent genes, -myosin heavy chain (MHC) and myosin-binding protein C, account for roughly 70% of all mutations. All mutations originate in genes that encode sarcomere proteins or are involved with sarcomere-related structures and are inherited in an autosomal dominant way. Genetic heterogeneity, like clinical diversity in illness, is a key feature of HCM. Many other disease genes have been linked to HCM, but these are more likely to be diseases that look like HCM but are caused by mutations in another gene, such as glycogen storage diseases caused by PRKAG2 mutations.

Two major techniques - linkage analysis and genetic association - have been used to find genes for CVD and associated risk factors in humans. The method chosen was determined by the pattern of segregation, which was either consistent with Mendel's ratios or more complex. Some types of CVD have a straightforward inheritance pattern that suggests a single causal gene with a big effect on phenotype. Direct DNA sequencing and/or linkage analysis have successfully identified the causal gene and mutation in several of these Mendelian types of CVD. Most CVD features, such as MI or plasma LDL cholesterol concentrations, have a complex inheritance pattern, indicating a complex interaction between numerous genes and non-genetic variables [1-3].

Although single gene changes can lead to simple genotype-phenotype correlations in some circumstances, there are also more complex relationships. Pleiotropy, penetrance, and expressivity are three different genetic phenomena that might cause this complexity. Mutations in a single gene can sometimes affect many phenotypic features (i.e., pleiotropy). Pleiotropy, penetrance, expressivity, and non-genetic variables all work together to ensure that genotype does not "equal" phenotype, even in single gene disorders. This complexity has a number of ramifications. First, gene discovery is more challenging since genotype and phenotype may not segregate correctly, limiting the power of linkage. Second, identifying modifiers - genetic or environmental - that may regulate the link between genotype and phenotype is of great interest [4,5].

Conclusion

Finally, due of this intricacy, developing genotype-specific prognostic or therapeutic recommendations for many Mendelian disorders has proven difficult. Patients with pathogenic Mitochondrial DNA (mtDNA) mutations are more likely to have cardiovascular problems. The link between CVD susceptibility and 'natural' mtDNA polymorphisms needs to be investigated further, however various new experimental models will help soon. Several genetic investigations in HCM have lately shown that up to 5% of families had two disease-causing gene variants. The ultimate therapeutic or biological significance of the gene identified by a genetic variant may have minimal link with the phenotypic variance explained by the variable. A variant's ability to explain phenotypic variance is determined by two essential parameters: allele frequency and effect size. Finally, the fusion of human genetics and functional biology will disclose new therapeutic targets for cardiovascular illness, as well as a new generation of drug development initiatives.

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