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Genetics of Dilated Cardiomyopathy: Clinical Implications.

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Abstract

PURPOSE OF REVIEW: This review aims to summarize the current knowledge on the genetic background of dilated cardiomyopathy (DCM), with particular attention to the genotype-phenotype correlations and the possible implications for clinical management.

RECENT FINDINGS: Next generation sequencing (NGS) has led to the identification of an increasing number of genes and mutations responsible for DCM. This genetic variability is probably related to the extreme heterogeneity of disease manifestation. Important findings have associated mutations of Lamin A/C (LMNA) and Filamin C (FLNC) to poor prognosis and the propensity to cause an arrhythmic phenotype, respectively. However, a deeper understanding of the genotype-phenotype correlation is necessary, because it could have several implications for the clinical management of the patients. Furthermore, the correct interpretation of pathogenicity of mutations and the clinical impact of genetic testing in DCM patients still represent important fields to be implemented. A pathogenic gene mutation can be identified in almost 40% of DCM patients. The recent discoveries and future research in the field of genotype-phenotype correlation may lead to a more personalized management of the mutation carriers towards the application of precision medicine in DCM.

KEYWORDS: Dilated cardiomyopathy; Filamin C; Genotype-phenotype correlation; Lamin A/C; Next generation sequencing; Precision medicine

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