Format: Abstract

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Genetics of Dilated Cardiomyopathy.

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Abstract

PURPOSE OF REVIEW: Dilated cardiomyopathy (DCM) is characterized by left ventricular dilation and systolic function and is the most common among all cardiomyopathies. Familial DCM makes up a significant portion of cases, and approximately 40 genes are identified as involved in the pathogenesis of heart failure, each affecting a specific part of cellular mechanisms. The purpose of this review is to summarize recent findings and the current understanding of the most common gene mutations identified associated with DCM.

RECENT FINDINGS: Next-generation sequencing is a comprehensive gene analysis technique used to discover more mutation variants and also to learn about the impact of mutations in relationship to clinical presentations. A variety of techniques are utilized to study different gene mutations, such as genotype-phenotype association analysis or whole-exome sequencing, to understand the natural history of diseases. For certain genetic abnormalities, information is helpful in developing potential therapeutic treatment targeting mutations. More treatment options are hopeful with the understanding of specific genetic mutations and their pathogenic mechanism. It also suggests the importance of genetic assessment and counseling for family members of an affected patient, in order to provide potential early diagnosis and better clinical management of DCM.

KEYWORDS: Dilated cardiomyopathy; Familial; Gene mutations; Lamin A/C; Screening; Titin

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