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Review Article

How genetic testing can be helpful in Dilated Cardiomyopathy: A Review

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Abstract

Characterised by ventricular chamber enlargement and systolic dysfunction, dilated cardiomyopathy (DCM) is a heterogeneous disorder with a strong predisposition for genetic mutations. In this review, we provide a synopsis on the etiological perspective of DCM paired with the part genetic testing plays in diagnostics, prophecy, and treatment before emphasising certain genetic tests available and their medically measurable implications.

1. Introduction

Systolic dysfunction and ventricular chamber enlargement are hallmarks of dilated cardiomyopathy (DCM). DCM is one of the primary drivers of heart failure and can be brought about by different variables, including hereditary transformations. The past decade has featured prominent strides in tracing the genetic basis associated with DCM - more than 50 genes being implicated - also making genetic examination integrally crucial during diagnosis, monitoring, and regulation within DCM care [1, 2].

1.1. Genetic Basis of DCM:

The genetic causes of DCM are complex. Mutations in genes encoding sarcomeric proteins, cytoskeletal proteins, nuclear envelope proteins, calcium-handling proteins and ion channels have been linked to the pathophysiology of DCM. The most common genetic cause of DCM is mutations in the gene encoding the sarcomeric protein titin (TTN)-the largest known human protein. 20-25% DCM patients have mutations in TTN [3, 4].

1.2. Role of Genetic Testing in DCM:

Genetic testing serves important diagnostic, prognostic, and treatment roles in DCM. For diagnosed DCM patients, genetic tests can help identify specific mutations causing the disease; thereby assisting in predicting disease progression, assessing risk, and guiding treatments. Additionally, asymptomatic relatives at risk for developing DCM (due to family history) can also benefit from genetic testing and prevention strategies can be employed that might ward off future complications [1, 5].

1.3. Specific Genetic Tests for DCM

Various specific genetic tests exist, which include the following approaches: Single gene testing - optimal for detecting known familial mutations with high suspicion rates; Panel testing suitable when investigators search multiple genes at lower suspicion indices; Whole exome sequencing (WES) - best utilised when there is a low index of suspicion and patients require comprehensive screening. Included below are some examples of genetic tests available used for diagnosing DCM: - TTN sequencing: This test involves sequencing the entire TTN protein-coding gene since it's prevalently associated with DCM [3]. Comprehensive panel testing: Another method is to employ panel testing with several implicated genes including LMNA, MYH7, and SCN5A among others [6, 7]. Mitochondrial DNA sequencing: In some scenarios, mutations in mitochondrial DNA could cause DCM; hence, researchers also consider full sequencing of the mitochondrial genome [8].

2. Conclusion

Genetic testing has become an important tool in the diagnosis and management of DCM. Advances in genetic testing technology have enabled rapid and comprehensive evaluation of the genetic basis of DCM. Genetic testing can provide important information about disease prognosis, response to therapy, and risk of disease development in asymptomatic relatives. Clinicians should consider genetic testing in all patients with DCM, and they should work.

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