Risk Stratification of Genetic, Dilated Cardiomyopathies Associated With Neuromuscular Disorders: Role of Cardiac Imaging.

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Abstract
The etiology of dilated cardiomyopathy (DCM) can be grouped as either genetic or nongenetic. More than 50 pathogenic genes have been described, with sarcomeric and lamin A/C mutations being the most common. Mutation carriers for genetic DCM are often asymptomatic until cardiac disease manifests with heart failure, arrhythmias, or sudden cardiac death. Preventive strategies are promising but can only be applied and tested adequately if genetic DCM can be diagnosed at an early stage. Early diagnosis of mutation carriers that may develop overt DCM requires advanced imaging techniques that can detect subtle structural and functional abnormalities. Advanced echocardiographic techniques such as tissue Doppler imaging and speckle tracking strain analysis permit early detection of functional abnormalities, whereas cardiovascular magnetic resonance techniques provide information on tissue characterization and myocardial energetics that may be altered at an early stage. Furthermore, nuclear imaging techniques provide information on cellular function (metabolism, perfusion). Once the diagnosis of overt DCM has been established, various imaging parameters such as echocardiography-based myocardial mechanics and cardiovascular magnetic resonance-based tissue characterization have shown incremental benefit to left ventricular ejection fraction in risk stratification. Further research is required to understand how imaging techniques may help to choose management strategies that could delay progression when instituted early in the course of the disease. The present article reviews the role of imaging in the risk stratification of genetic DCM in general, with specific emphasis on DCM associated with neuromuscular disorders.

KEYWORDS: diagnosis; dilated cardiomyopathy; genetic; imaging techniques; risk stratification

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