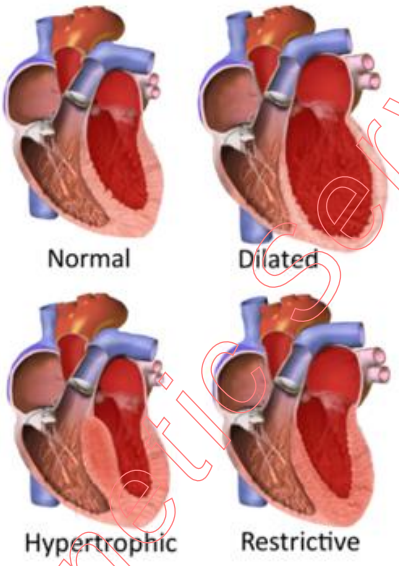
Genetic Science Spotlight

Northwestern University: Molecular Diagnostics for Cardiomyopathy Aids Precision Medicine



CENTER FOR GENETIC MEDICINE

Most of cardiomyopathy cases have a genetic origin. With the advances in genetic technologies, nearly 100 genes have been identified as associated with different types of cardiomyopathies, especially with dilated cardiomyopathy (DCM) and hypertrophic cardiomyopathy (HCM). The large number of genes responsible for cardiomyopathy, as well as the myriad of diverse mutations within each of these genes, produces remarkable heterogeneity for this complex disorder. Individual cardiomyopathy-associated genetic variants are infrequent in the general population (< 1 in 500), and individual genetic variants associate with a range of expressivity causing mild and severe forms of disease. Despite the complexity in genotype-phenotype association, researchers have successful relate several key genes and pathways associated with the condition. HCM has observed genetic differentiation that is clinically meaningful in terms of cardiovascular physiology, and the same classification will develop for DCM. In addition to being a tool for early diagnosis, risk assessment, prognostic stratification and, possibly, adoption of primary preventive measures in affected patients and their asymptomatic relatives, molecular diagnostic tests for cardiomyopathy may also be a strong advocate of precision medicine by classifying cardiomyopathy subtypes.

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4331062/

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