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Front Physiol. 2018 Sep 25;9:1356. doi: 10.3389/fphys.2018.01356. eCollection 2

Role of Lamin A/C Gene Mutations in the Signaling Defects Leading to Cardiomyopathies.

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Abstract

Nuclear lamin A/C are crucial components of the intricate protein mesh that underlies the inner nuclear membrane and confers mainly nuclear and cytosolic rigidity. However, throughout the years a number of other key physiological processes have been associated with lamins such as modulation of both genes expression and the activity of signaling mediators. To further solidify its importance in cell physiology, mutations in the lamin A/C gene (*LMNA*) have been associated to diverse pathological phenotypes with skeletal muscles and the heart being the most affected systems. When affected, the heart develops a wide array of phenotypes spanning from dilated cardiomyopathy with conduction defects to arrhythmogenic right ventricular cardiomyopathy. The surprising large number of cardiac phenotypes reflects the equally large number of specific mutations identified in the *LMNA* gene. In this review, we underlie how mutations in *LMNA* can impact the activity and the spatial/temporal organization of signaling mediators and transcription factors. We analyzed the ever-increasing amount of findings collected in $Lmna^{H222P/H222P}$ mice whose cardiomyopathy resemble the most important features of the disease in humans and a number of key evidences from other experimental models. With this mini review, we attempt to combine the newest insights regarding both the pathogenic effects of *LMNA* mutations in terms of signaling abnormalities and cardiac laminopathies.

KEYWORDS: Ca2+ signaling; cardiac pathophysiology; gene expression; lamin A/C gene; nuclear envelope; nuclear lamina; nucleus; signaling pathways

PMID: 30319452 PMCID: PMC6167438 DOI: 10.3389/fphys.2018.01356

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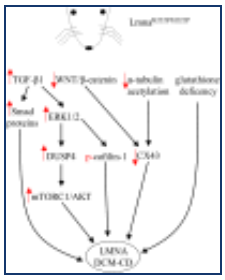

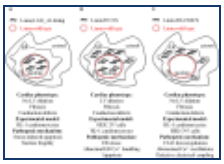

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