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## Effect of SCN5A Gene Mutation in Endocardial Cell

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**Abstract :** The simulation of an endocardial cell for gene mutation in the cardiac sodium ion channel NaV1.5, encoded by SCN5A gene, is discussed. The characterization of Brugada Syndrome by loss of function effect on SCN5A mutation due to L812Q mutant present in the DII-S4 transmembrane region of the NaV1.5 channel protein and its effect in an endocardial cell is studied. Ten Tusscher model of human ventricular action potential is modified to incorporate the changes contributed by L812Q mutant in the endocardial cells. Results show that BrS-associated SCN5A mutation causes reduction in the inward sodium current by modifications in the channel gating dynamics such as delayed activation, enhanced inactivation, and slowed recovery from inactivation in the endocardial cell. A decrease in the inward sodium current was also observed, which affects depolarization phase (Phase 0) that leads to reduction in the spike amplitude of the cardiac action potential.

**Keywords:** SCN5A gene mutation, sodium channel, Brugada syndrome, cardiac arrhythmia, action potential **Conference Title:** ICBMCE 2020: International Conference on Biological, Medical and Chemical Engineering

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