

CACNA1C Timothy syndrome

<https://pubmed.ncbi.nlm.nih.gov/32629111/>

L-type voltage-gated calcium channels play an essential role in various physiological systems including neuronal excitation and any mutation or dysfunction in the channel has significant impact on human brain function resulting in psychiatric diseases. Particular gain-of-function mutations in CACNA1C encoding Ca

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1.2 have been associated with Timothy Syndrome, a devastating disease with a multi-organ phenotype. Efforts to understand the underlying pathophysiology and find therapeutic strategy have been spurred recently with the advances in stem cell technology, in particular those arising from patient-derived sources. In this review, we report on the recent advances in Timothy Syndrome research and on the methods used to study this disease.