CDB SEMINAR


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16:00~17:00 A7F Seminar Room

# Using Patient Derived iPS Cells to Investigate Cellular Phenotypes in Timothy Syndrome


**Summary**

Timothy syndrome is a disorder characterized by multi-organ dysfunctions including long QT syndrome, cardiac arrhythmia and autism. Although the *de novo* missense mutation in L-type calcium channel ($Ca_V1.2$) causes Timothy syndrome, how the mutation alters calcium signaling and electrical activity in human hearts and brains remains unknown. Mouse models of human cardiac arrhythmia have proved to be problematic because the mouse resting heart rate is approximately tenfold faster than that of humans and therefore mouse cardiomyocytes have different electrical properties from their human counterparts. Furthermore, there are obvious differences in the development and function of brains between human and rodents. Taking advantage of human induced pluripotent stem (iPS) cells, we generated human cardiomyocytes and neurons to investigate cellular phenotypes in Timothy syndrome using a variety of cellular assays including live cell imaging, electrophysiological recordings, gene expression profiling and immunocytochemistry. Here I report the phenotypes that were observed in cardiomyocytes and neurons derived from Timothy syndrome patients. In addition, I talk about future directions of researches using human iPS cells as well as a directed test of chemical compounds and blockers to rescue the cellular defects in Timothy syndrome.



Keyword:
Patient derived iPS cells, Timothy syndrome, calcium signaling, cardiac arrhythmia, autism & drug screen



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