

---

---

# The 71<sup>st</sup> iCeMS SEMINAR

---

---

2011 3. 8 (火)

16:00-17:00

演 題 : **Using Patient Derived iPS Cells to Investigate Cellular Phenotypes in Timothy Syndrome**

講演者 : 矢澤 真幸 博士

米国 スタンフォード大学医学部神経学科

場 所 : 京都大学 アイセムス本館 2階 (東一条北西角)  
セミナー室 (A207)

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 (CaV1.2) 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.

主 催 : 京都大学 物質-細胞統合システム拠点 (iCeMS = アイセムス)

連絡先 : iCeMS イノベーションマネジメントグループ e-mail: sengoku-g@icems.kyoto-u.ac.jp

