Disease Characterization of Long QT Syndrome Using iPS Cell-Derived Cardiomyocytes

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Long QT syndrome (LQTS) is well characterized inheritable, life threatening disease, and often related to large pedigrees of family history. The present study established induced pluripotent stem cell (iPSC) from the patients with LQTS, and investigated whether the LQTS patient-derived iPSCs can be utilized for disease characterization, and drug response. We reprogrammed patient somatic cells, differentiated cardiomyocytes and examined electrophysiological properties. Genotype analysis showed the heterozygote mutation in KCNQ1 gene, 1893delC, indicating that this patient was type 1 LQTS. Drug response examination using multi-electrode analysis (MEA) revealed the LQTS-iPSC-derived cardiomyocytes revealed IKs disturbance, but not IKr. Electrophysiological recording confirmed 1893delC has a dominant negative role in IKs channel function by trafficking defect. Isoproterenol induced ventricular tachycardia-like arrhythmia. This study provides the evidences that iPSCs can be utilized for characterization, drug response, and diagnosis for patients with LQTS to conduct medical therapies.

Keywords: iPSC cell, Long QT syndrome, sudden cardiac death