STEMOD™ LQT1 Disease Model (KCNQ1 (KVLQT1, Kv7.1), R190Q (G569A))

Cat. No.: MoCVD-ZXY001

This product is for research use only and is not intended for diagnostic use.

PRODUCT INFORMATION

Product Overview

Long-QT syndromes are heritable diseases associated with prolongation of the QT interval on an electrocardiogram and a high risk of sudden cardiac death due to ventricular tachyarrhythmia. This disease cell model belongs to long-QT syndrome type 1, an autosomal dominant missense mutation (R190Q) occur in the KCNQ1 gene. Generating pluripotent stem cells from dermal fibroblasts, then differentiate into cardiac myocytes. This disease cell model provides a platform to explore disease mechanisms in human genetic cardiac disorders.

SPECIFICATIONS

Organ System

Cardiovascular System

Disease

LQT1

Target Gene

KCNQ1 (KVLQT1, Kv7.1)

Gene Function

KCNQ1 encodes the repolarizing potassium channel mediating the delayed rectifier Iks current, pore-forming α subunit of the channel generating the slow component of the delayed rectifier potassium current Iks

Mutation

R190Q (G569A)

Phenotype

Dominant-negative trafficking defect, reduced Iks

Source

Dermal fibroblasts

Cellular Assays

Whole-cell patch clamp techniques, immunohisto-chemistry
## GENE INFORMATION

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