STEMOD™ LQT3/BrS Disease Model (SNC5A, N406K (1218C>A))

Cat. No.: MoCVD-ZXY020

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 3, an mutation (N406K, also called 1218C>A) occur in the SNC5A gene. Generating pluripotent stem cells from skin 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
LQT3/BrS

Target Gene
SNC5A

Gene Function
SNC5A encodes the α-subunit of the cardiac sodium channel, Nav1.5, and the mutation lead to the sodium current (INa) failing to inactivate properly, thereby increasing APD and prolonging cardiomyocyte repolarisation.

Mutation
N406K (1218C>A)

Phenotype
Prolonged action potential duration (APD) and Ca2+ transients, EADs

Source
Skin fibroblasts

Cellular Assays
Whole-cell patch clamp techniques
GENE INFORMATION

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