STEMOD™ LQT3/BrS Disease Model (SNC5A, p.V1763M (c.5287G> A))

Cat. No.: MoCVD-ZXY019

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 (p.V1763M, also called c.5287G> A) occur in the SNC5A 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 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 p.V1763M (c.5287G> A)
Phenotype Faster recovery from INa inactivation, increased INa, prolonged action potential duration (APD)
Source Dermal fibroblasts
Cellular Assays Whole-cell patch clamp techniques, immunostaining assay
GENE INFORMATION

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