**STEMOD™ LQT1/JLNS Disease Model (KCNQ1 (KVLQT1, Kv7.1), R594Q (c.1781G>A))**

Cat. No.: MoCVD-ZXY007

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

**PRODUCT INFORMATION**

**Product Overview**

Jervell and Lange-Nielsen syndrome (JLNS) is one of the most severe life-threatening cardiac arrhythmias. This disease cell model belongs to long-QT syndrome type 1, mutation (R594Q, also called c.1781G>A) 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/JLNS
- **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**: R594Q (c.1781G>A)
- **Phenotype**: Prolonged action potential duration (APD), trafficking defect
- **Source**: Dermal fibroblasts
- **Cellular Assays**: Whole-cell patch clamp techniques, immunostaining
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