

**Technology**

Genetic testing for diagnosis and/or prognosis of cardiac arrhythmia

**Inventor**

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**Key Features**

- Sequenced approximately 100kb of the *SCN5A* promoter of the human voltage-gated sodium channel
- Potential to improve arrhythmia-specific genetic testing
- Potential to impact cardiac arrhythmia diagnosis and treatment

**Stage of Development**

Reduced to practice with successful demonstration in vitro studies

**Keywords**

Diagnostic  
- Genetic testing  
- Cardiac Arrhythmia

**Patent Status**

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## Human Sodium Channel Promoter Region (VA Reference No. 05-151)

*Useful tool in the development of human genetic testing for sudden death risk associated with cardiac arrhythmia*

**Technology**

The Department of Veterans Affairs has identified a full, complete description of the promoter region and the mRNA untranslated regions of the human cardiac sodium channel for fetal and adult hearts. Many mutations or dysregulations of this channel cause human, inherited sudden death syndromes. The knowledge demonstrated will likely prove useful in future human genetic testing for sudden death risk.

**Description**

Mutations within the *SCN5A* gene are responsible for a number of cardiac arrhythmias, including Long QT-3 (LQT3), Brugada Syndrome and isolated cardiac conduction disease (ICCD). The developed technology represents a useful tool for future genetic testing for *SCN5A*-linked cardiac arrhythmias. Previous research has focused on the coding regions of *SCN5A*, but researchers from the VA focused their analysis on the promoter and 5' and 3' untranslated regions of the *SCN5A* gene. Alterations within the promoter region could potentially alter the expression of the *SCN5A* protein, leading to an arrhythmic state. Significantly, multiple 5' and 3' mRNA splice variants have also been identified. These could encode *SCN5A* isoforms possessing different, perhaps cardiac disease-associated, biological activities. Thus, these findings could potentially impact genetic testing and treatment for *SCN5A*-linked arrhythmias

**Competitive Advantage**

Unlike current methods of diagnosis and/or prognosis for cardiac arrhythmia, this invention:

- Has the potential to impact cardiac arrhythmia treatments by identifying patients with an arrhythmia-linked genetic mutation.
- Has the potential to improve arrhythmia-specific genetic testing leading to enhanced diagnosis and therapy.

**Status**

The Department of Veterans Affairs is looking for a partner for further development and commercialization of this technology through a license, and the VA inventors are available to collaborate with interested companies through a Cooperative Research and Development Agreement (CRADA).