

Analysis of Cardiac Ion Channels to Understand Arrhythmias Which Lead to Sudden Cardiac Death

Authors: Carbone, S.L.; Clark, C.S.; Entelisano, C.M.; Barlett, M.A.; Miller, A.L.

Mohawk Valley Community College, Utica, NY

Betzenhauser, M.J.

Masonic Medical Research Laboratory, Utica, NY

There are 300,000-400,000 fatalities attributed to sudden cardiac death every year in the U.S. due to a lack of sufficient research on mechanisms causing arrhythmias¹. Malfunctions with the ion channels in the heart may lead to lethal arrhythmias. The purpose of this work is to study ion channels and evaluate malfunctions relative to normally functioning hearts. Plasmid insertion in *E. coli* assayed whether functional ion channels reach the membrane, and confocal fluorescent microscopy was used to illuminate cellular functionality. In addition, genetic analysis was used to determine the extent of hereditary factors in sudden cardiac death. Genes that encode for the voltage-gated sodium, potassium, and calcium ion channels were analyzed at the genetic level using isolated DNA samples and traditional Sanger sequencing methods to identify mutations that may be responsible for sudden cardiac death syndromes. For example, Long QT syndrome, Short QT syndrome, and Brugada syndrome are caused by mutations in these ion channels. Once these mutations are identified, genetic engineering techniques can be used in the generation of new heart cells from the stem cells found in somatic tissue. Generation of such heart cells is important because it could lead to the development of personalized treatment for degenerative diseases such as heart failure in the future.

1. Rubart, M. et al., *Mechanisms of Sudden Cardiac Death*, **2005**. J. clin. invest. 115(9):2305-2315.