ABSTRACT

The field of molecular arrhythmology has progressed at an impressive pace during the past 20 years. Throughout the years, we have learned more and more about the genetic factors and molecular mechanisms underlying electrical abnormalities of the heart such as congenital long QT syndrome (LQTS) and Brugada syndrome. Since in most cases, the genes that are found to be mutated are encoding either the pore-forming subunit of cardiac ion channels or of ion channel regulatory proteins, the term “genetic cardiac channelopathies” has been used to define these disorders. Among the still-growing list of genes that lead to genetic cardiac channelopathies, the role of the gene, SCN5A, is truly unique. The gene SCN5A encodes the pore-forming subunit of the cardiac sodium channel, Nav1.5, which is the main channel responsible for the cardiac sodium current. Hundreds of genetic variants have been reported. The striking point here is that these variants were found in patients with a long list of distinct clinical manifestations ranging from delayed repolarization (in LQTS) to structural abnormalities (in the case of patients with dilated cardiomyopathies). During this research seminar, new findings related to the roles and regulation of Nav1.5 in cardiac cells will be presented. In particular, most of the recent results are coming from the investigation of genetically-modified mouse models.

BIOGRAPHY

Hugues Abriel studied life sciences at the Swiss Federal Institute of Technology in Zurich (ETHZ, 1989). He continued his education to become a physician (MD, 1994) and received a PhD degree in Physiology from the University of Lausanne in Switzerland (1995). He has spent two years as a research scientist at Columbia University in New York, USA. Hugues Abriel has been a group leader (2002-2009) at the Department of Pharmacology and Toxicology at the University of Lausanne thanks to a professorship from the Swiss National Science Foundation (SNSF-Professor). Since 2009, he is the Director of the Department of Clinical Research of the University of Bern at the Inselspital, and professor of pathophysiology. In 2012, he was elected as a member of the research council of the Swiss National Science Foundation. He is since 2015 the Director of the National Center of Competence in Research TransCure, which is a network of Swiss scientist who want to apply excellence in membrane transporter research to the treatment of human diseases. His research work focuses on the roles of ion channels in human diseases (channelopathies). Currently, he is mainly exploring the genetic, molecular and cellular bases of cardiac arrhythmias.