PIONEERS IN BIOMEDICAL RESEARCH SEMINAR

Presented by the Fralin Biomedical Research Institute at VTC and co-sponsored by the institute's Center for Vascular and Heart Research



MARIO DELMAR, M.D., Ph.D.

Endowed Professor of Medicine Patricia and Robert Martisen Professor of Cardiology Leon H. Charney Division of Cardiology New York University School of Medicine

Sudden Cardiac Arrest in the Young: The Case of ARVC

Plakophilin-2 (PKP2) is a protein of the desmosome, an intercellular adhesion structure. Pathogenic variants in the gene coding for PKP2 associate with most cases of gene-positive arrhythmogenic right ventricular cardiomyopathy (ARVC), a disease characterized by the loss of muscle mass at the expense of fibrofatty infiltrates (predominantly in the right ventricle), ventricular arrhythmias, and high propensity for sudden death in the young. Importantly, exercise in PKP2 pathogenic variant carriers significantly increases the risk of developing the cardiomyopathy, accelerates the progression to heart failure, and increases the occurrence of arrhythmias and sudden death. Current ARVC therapy is not curative, and only mildly effective in alleviating symptoms and containing disease progression. The long-term goals of Dr. Delmar's laboratory are to advance our understanding of ARVC molecular mechanisms, and to generate pre-clinical knowledge that can improve ARVC therapy and evaluation of risk. In this seminar, Dr. Delmar will present his latest findings on the molecular underpins of the ARVC cardiac phenotype, with particular emphasis on exercise-induced arrhythmogenic cardiomyopathy. He will also present his lab's recent preclinical data on AAV-mediated gene replacement therapy, which provided the groundwork for an ongoing clinical trial on gene therapy to cure PKP2-dependent ARVC.

FRIDAY, OCT. 18, at 11 a.m.

Room G101 A/B, 4 Riverside Circle Watch live via Zoom at <u>https://FralinBioMed.info/PBR-Join</u>

