A white male patient aged 52 years with permanent atrial fibrillation and left ventricular hypertrophy because of glycogen storage cardiomyopathy, heterozygous for c.905G>A p.(Arg302Gln) PRKAG2 missense mutation on exon 7, was admitted because of partial pacemaker extrusion and pocket infection. His mother and 5 brothers carry the same mutation and were previously reported. A dual chamber pacemaker was implanted 20 years earlier because of third-degree atrioventricular block. Pacemaker leads and generator were explanted under extracorporeal circulation. A fragment from the base of the right atrial appendage was assessed for microscopic analysis. Percutaneous endomyocardial right ventricle (RV) biopsy was undertaken 1 year before. Morphological changes in ventricular myocardium in PRKAG2 cardiomyopathy have been reported, but there are no data on atrium pathology. Atrial section (Figure A through C) stained with periodic acid–Schiff (Figure A) and hematoxylin and eosin (Figure B) shows intense vacuolization of the myofibers with abundant gross granular inclusions—glycogen—(arrows) within vacuoles. There is no cardiomyocyte architecture disarray. Masson trichrome staining (Figure C and F) shows normal collagen fibers (blue) in the extracellular matrix, without fibrosis. Lower (Figure D through F) show histopathology findings in the RV, which were similar to those in the atrium, with vacuolization, absence of myocardial disarray, inflammatory cells, and fibrosis, findings consistent with RV histopathology previously reported. Transmission electron microscopy image from RV septum (D) shows mitochondrial within abundant glycogen (small granules), between myofibrils.

Patients with R302Q have a high incidence of atrial fibrillation. Ventricular preexcitation and hypertrophy, which are commonly present in these patients, may be contributors. It has also been suggested that in the absence of fibrosis, a reduction of pH because of increased glycogen content could influence ionic channels function, which may contribute to atrial fibrillation maintenance. Further studies are warranted in this model of persistent atrial fibrillation without fibrosis.

Disclosures
None.

References

Key Words: atrial appendage • atrial fibrillation • collagen • glycogen storage cardiomyopathy • PRKAG2 mutation
Atrial Histopathology in PRKAG2 Cardiomyopathy

Figure. Atrial section (A–C) stained with periodic acid–Schiff (A) and hematoxylin and eosin (B) shows intense vacuolization of the myofibers with abundant gross granular inclusions—glycogen—(arrows) within vacuoles. There is no cardiomyocyte architecture disarray. Masson trichrome staining (C and F) shows normal collagen fibers (blue) in the extracellular matrix, without fibrosis. D to F, lower, Show histopathology findings in the right ventricle (RV), which were similar to those in the atrium, with vacuolization, absence of myocardial disarray, inflammatory cells, and fibrosis, findings consistent with RV histopathology. Transmission electron microscopy image from RV septum (D) shows mitochondria (M) within abundant glycogen (small granules), between myofibrils (MF).
Atrial Pathology Findings in a Patient With PRKAG2 Cardiomyopathy and Persistent Atrial Fibrillation
Eduardo Back Sternick, Stanley de Almeida Araújo, Elizabeth Ribeiro da Silva Camargos and Geraldo Brasileiro Filho

Circ Arrhythm Electrophysiol. 2016;9:
doi: 10.1161/CIRCEP.116.004455
Circulation: Arrhythmia and Electrophysiology is published by the American Heart Association, 7272 Greenville Avenue, Dallas, TX 75231
Copyright © 2016 American Heart Association, Inc. All rights reserved.
Print ISSN: 1941-3149. Online ISSN: 1941-3084

The online version of this article, along with updated information and services, is located on the World Wide Web at:
http://circep.ahajournals.org/content/9/12/e004455

Permissions: Requests for permissions to reproduce figures, tables, or portions of articles originally published in Circulation: Arrhythmia and Electrophysiology can be obtained via RightsLink, a service of the Copyright Clearance Center, not the Editorial Office. Once the online version of the published article for which permission is being requested is located, click Request Permissions in the middle column of the Web page under Services. Further information about this process is available in the Permissions and Rights Question and Answer document.

Reprints: Information about reprints can be found online at:
http://www.lww.com/reprints

Subscriptions: Information about subscribing to Circulation: Arrhythmia and Electrophysiology is online at:
http://circep.ahajournals.org/subscriptions/