Images in Cardiovascular Medicine

Association of Left Ventricular Noncompaction with Polycystic Kidney Disease

as Shown by Cardiac Magnetic Resonance Imaging

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A 37-year-old man with a history of polycystic kidney disease (PCKD) was admitted with new-onset congestive heart failure. In regard to the patient's family history, his father also had a history of PCKD, before dying at age 50 years with an undocumented "heart problem." Echocardiograms showed a left ventricular (LV) ejection fraction of <0.20. Coronary angiography showed normal coronary arteries with an LV end-diastolic pressure of 30 mmHg.

Cardiac magnetic resonance (CMR) images showed hypertrabeculation along the LV apex and lateral wall, with reduced thickness of the compacted layer. The CMR images, including those of the upper abdomen, showed multiple bilateral cysts (Figs. 1–3). At the time of publication, the patient was being considered for an implantable cardioverter-defibrillator for primary prevention, and also for later cardiac transplantation. Genetic testing had not been performed, for financial reasons.

Comment

Left ventricular noncompaction (LVNC) is a rare abnormality of cardiac development in which there is failure of the endocardium to properly organize, secondary to intrauterine arrest of the myocardial compaction process. This condition displays a wide spectrum of genetic penetrance and symptomatic severity. Often it is confined to the LV apex or to the mid-distal inferior and inferolateral walls.[1]

Left ventricular noncompaction has been associated with the formation of multiple cysts of the kidneys, as in our patient, and with hepatic, pancreatic, and intracranial aneurysms.[2] The *PCKD1* and *PCKD2* genes, responsible for the synthesis of polycystin, have a role in the onset of autosomal dominant PCKD. The deletion of *PCKD1*

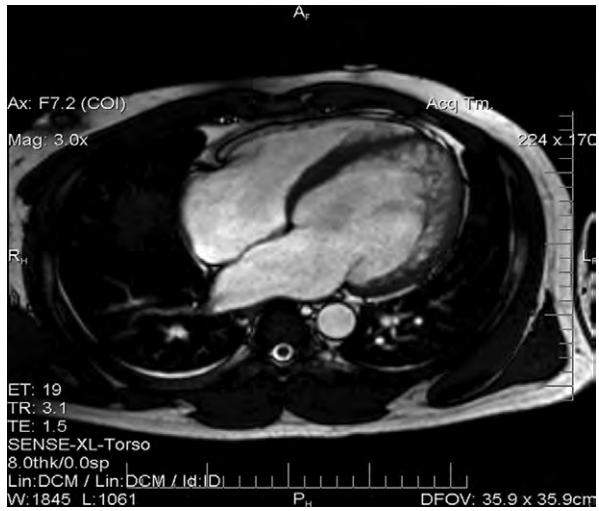

Fig. 1 *Cardiac magnetic resonance image from an axial steady-state free-precession sequence, obtained at the level of the left ventricle, shows noncompaction changes within the myocardium of the left ventricle.*

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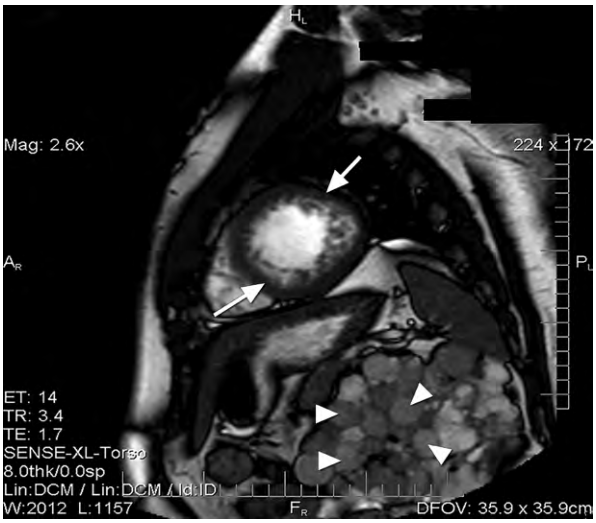

Fig. 2 *Cardiac magnetic resonance image from a sagittal steady-state free-precession sequence, obtained at the level of the left ventricle (including the upper abdomen), shows noncompaction of the left ventricular myocardium (arrows) and, in the enlarged left kidney, multiple cysts (arrowheads) with various signal intensities.*

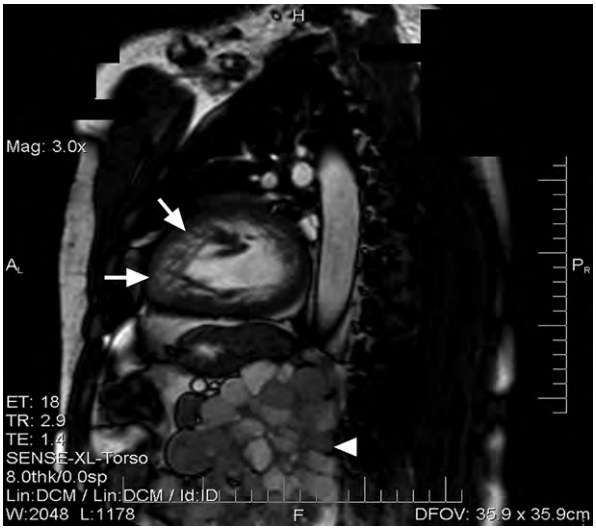

Fig. 3 *Cardiac magnetic resonance image from an oblique sagittal steady-state free-precession sequence, obtained at the level of the left ventricle, shows noncompaction of the left ventricular myocardium (arrows). Upper abdomen shows multiple and various cystic changes in the left kidney (arrowhead).*

or *PCKD2* is responsible for PCKD, which is linked to disorganized myocardial arrangement in mice with such deletions.[3]

To the best of our knowledge, ours is only the 3rd adult case reported in the literature with this apparent association between PCKD and LVNC.[4,5] This association in human beings might be "speculative," but there remains the genetic link between PCKD and LVNC in lower animals.[3]

We are safe to assume that, in a patient with PCKD, a cardiac diagnostic evaluation would most often consist only of an echocardiogram, which can miss LVNC because of the patient's lack of global LV involvement, echocardiography's inability in many instances to fully render the LV apex, and the examining physician's lack of familiarity with LVNC. Added to these diagnostic impediments is the fact that very heavily trabeculated ventricles frequently lead to overcalling LVNC on the basis of morphologic imaging criteria alone. Not all patients with PCKD undergo cardiac imaging that is adequate to fully exclude LVNC. Therefore, LVNC is frequently both "overcalled" and "undercalled."

When a patient with PCKD is encountered, prospective studies that use CMR imaging (the gold standard) or very carefully directed echocardiography can be useful in establishing the extent of association between LVNC and PCKD. Left ventricular noncompaction presents as a spectrum: many patients with regional disease are treated conservatively, with an apparently low risk for bad outcome. However, heightened suspicion of concomitant cardiomyopathy might be warranted to prevent sudden cardiac death by instituting targeted cardioprotective therapy in a timely fashion.[6]

Although LVNC is more commonly seen now that cardiac multimodality imaging is so widespread, concomitant PCKD is not present in the vast majority of LVNC cases—even the most dramatic. That said, the frequency of LVNC in association with PCKD might well be underreported; it certainly deserves further study.

References

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