A rare cause of cardiomyopathy: Left ventricular noncompaction cardiomyopathy

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Introduction

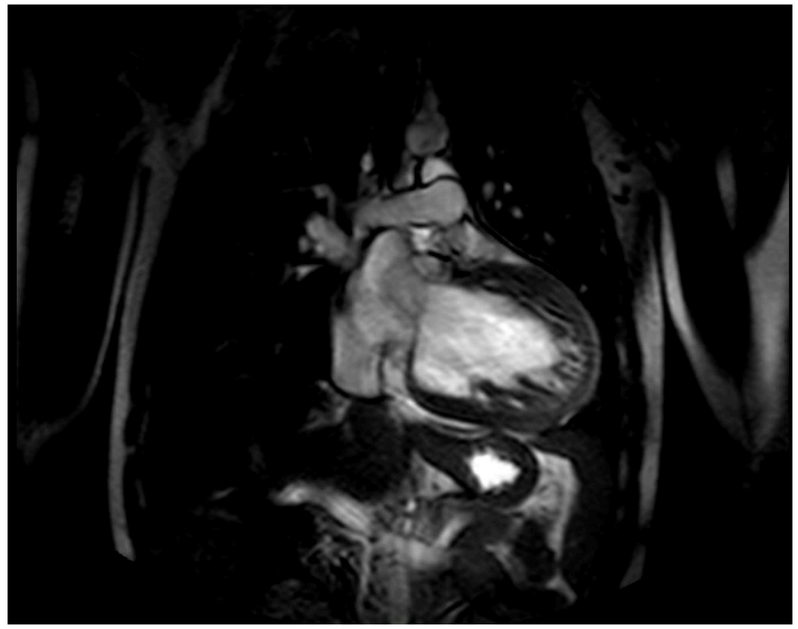
Cardiomyopathy can be divided into dilated, restrictive, hypertrophic and arrhythmogenic right ventricular cardiomyopathy [1]. We present a case of dilated cardiomyopathy due to rare cause: left ventricular noncompaction cardiomyopathy (LVNC).

Case discussion

34-year-old male who presents with chest pain associated with progressive, exertional dyspnea. Medical history includes previous use of oral opioids and current alcohol use disorder (4-5 shots of whisky per night), tobacco use and occasional marijuana use. Family history is significant for sudden cardiac death in father and paternal grandfather in the 40s and siblings diagnosed with Marfan's syndrome; however, he himself was tested negative for Marfan's syndrome. Vitals were significant for blood pressure of 170/71 and heart rate of 56. Physical examination was without any abnormal findings. Blood work including troponins were largely unremarkable. ECG showed sinus rhythm with a left bundle branch block (LBBB) pattern with occasional premature ventricular complexes. A transthoracic echocardiogram (TTE) was done which showed severely reduced left ventricular ejection fraction (LVEF) of 10-15% with global hypokinesis. It was also concerning for severe LV hypertrophy, prominent trabeculations in the LV apex and grade 2 diastolic dysfunction.

Cardiology was consulted. Patient underwent left heart catheterization to rule out ischemic cardiomyopathy. He was started on a small dose of furosemide and losartan. Beta blockade was not initiated due to bradycardia. Cardiac MRI (CMR) revealed a LVEF of 18%, global hypokinesis and prominent trabeculae in the LV raising the possibility of LVNC. Due to continued bradycardia and LBBB with intermittent 2:1 heart block, a conductive heart disease was suspected, and patient had a biventricular implantable cardioverter defibrillator (BiV-ICD) implanted. Beta blockade was initiated. Genetic test showed heterozygous pathogenic mutation in the CBS gene. The patient improved and was discharged and counselled about excessive alcohol use and the importance of following up with cardiology.

Imaging



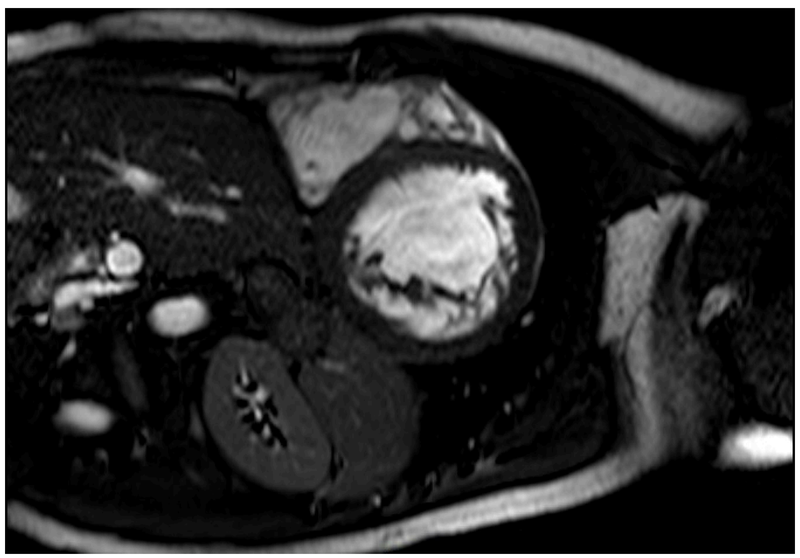


Figure 1 and 2: Cardiac MRI in different views showing prominent trabeculations in the left ventricle.

Discussion

LVNC is an unclassified, rare, and congenital cause of cardiomyopathy. It is associated with trabeculations and pockets within the myocardium that communicate with the LV. It is due to the inability of the spongy myocardium, during embryonic development, to become compacted during gestation, due to unknown reasons [2]. Heart failure, ventricular arrhythmias and embolism are some of the complications. Diagnoses is based on imaging with TTE showing non-compacted and compacted bilayered myocardium, prominent myocardium and deep endomyocardial recesses, whereas CMR evaluates different parameters involving noncompacted and compacted myocardium; however, no definite criteria has been validated [3]. Genetic testing should be carried out. There is no specific therapy and guideline directed therapy should be adopted to treat heart failure, including medical therapy and cardiac resynchronization therapy, if needed. The presence of heart failure, arrhythmias, and family history of premature death from heart disease as well as imaging findings should raise the suspicion for LVNC.

References

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