A 39-year-old woman visited an outpatient clinic complaining of nausea and vomiting for 1 week. She had been experiencing dyspepsia for several months. Her endoscopic gastroduodenography did not show any remarkable findings. She was lean and of short stature, with a weight of 29 kg, height of 147 cm, and an approximated body mass index of 13.49 kg/m². She had some hearing difficulties and cognitive dysfunction. Her parents died when she was a teenager; her mother passed away because of diabetic complications in her third decade. She was a nonsmoker and denied any history of hypertension or symptoms of chest pain except for alleged diabetes mellitus. Her chest x-ray film showed cardiomegaly, and ECG revealed normal sinus rhythm and left ventricular hypertrophy with T-wave inversion along V₄₋₆ precordial lead. Her blood troponin-T and creatine kinase MB levels were mildly elevated. Lactate dehydrogenase and B-type natriuretic peptide levels were also elevated to 456 IU/L and 147.0 pg/mL, respectively. Her hemoglobin A1C level was elevated, whereas Islet cell antibody and insulin antibody levels were normal, suggesting type 2 diabetes mellitus. On pure tone audiometry, sensory neural hearing loss at 2 kHz was noted in both ears: 70 dB in the right ear and 50 dB in the left. Transthoracic echocardiogram showed normal-sized cardiac chambers and severely reduced global left ventricular systolic function, with an estimated ejection fraction of 35% and a small amount of pericardial effusion. The left ventricular wall was thickened, and grade-2 diastolic dysfunction was noted (Figure 1). To characterize the myocardiun, cardiac MRI was performed, revealing a diffusely increased T2 value of 68 ms on T2 map. However, no late gadolinium enhancement (LGE) was noted. The major role of cardiac MRI is to exclude additional information on myocardial tissue status in a case differentiating from MC. Genetic study and systemic involvement are important for the diagnosis of MC. Findings of no infiltrative cardiomyopathy or hypertrophic cardiomyopathy can be helpful for obtaining information on myocardial tissue characteristics in MC.1,2 In the present study, T1, T2 mapping, and extracellular volume fraction analysis provided additional information on myocardial tissue status in a case without LGE. The major role of cardiac MRI is to exclude any infiltrative cardiomyopathy or hypertrophic cardiomyopathy in cases with thickened myocardium. In this case, after echocardiography, Fabry disease, cardiac amyloidosis, or diffusely thickened hypertrophic cardiomyopathy should be differentiated from MC. Genetic study and systemic involvement are important for the diagnosis of MC. Findings of no patch LGE in septa and no subendocardial LGE combined with a diffuse increase in T2 suggested a lower possibility of hypertrophic cardiomyopathy or cardiac amyloidosis.
because T2 increase is not observed in these disease entities. No native T1 shortening was a clue to exclude Fabry disease and other glycogen storage disease. T2-weighted images are now challenged by several limitations. To overcome these limitations, direct quantification of T1 and T2 through a mapping technique is helpful. The patient described herein exhibited m.3243A>G mtDNA mutation, which can cause MELAS syndrome, in which T2 has been reported to be increased. The T2 increase can be from myocardial edema at some degree or intracytoplasmic vacuole. Not only was clinically viral myocarditis not indicated in this case, but also the absence of subepicardial or midwall LGE in the absence of severe lymphocyte infiltration excluded acute or chronic myocarditis. Accordingly, such findings can provide clues to diagnosing MC.

Disclosures

None.

References


Figure 1. Parasternal long-axis (A) and short-axis (B) transthoracic echocardiography view shows thickened left ventricular myocardium with small pericardial effusion. Mitral inflow Doppler pattern (C) and tissue Doppler of mitral annulus and reversal of systolic and diastolic inflow pattern of pulmonic vein Doppler (D) suggest grade-2 diastolic dysfunction. PV indicates pulmonic vein.
Figure 2. Cardiac MRI findings. No late gadolinium enhancement (A) is noted. Measured T2 value in T2 map is increased by 68 ms (B). Precontrast (C) and postcontrast (D) T1 value in T1 map is 1078 ms and 469 ms, respectively, and then calculated extracellular volume fraction is increased by 34%. Region of interest placed on the entire myocardium.

Figure 3. Histopathologic findings of cardiac biopsy. A, Vacuolar change in myocardial cells and perimysial fibrosis, Hematoxylin and eosin, ×200. B, Gomori trichrome stain showing ragged red fibers, ×1000. C, Electron microscopy showing abnormal mitochondria with fingerprint arrangement, ×50,000.

Figure 4. Mitochondrial DNA PCR sequencing and m.3243A>G mutation.
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