A 46-year-old man presented with exertional dyspnea that had gradually progressed for 2 years. He had a 10-year history of hearing impairment. There was no other medical history including hypertension, diabetes mellitus, coronary artery disease, or arrhythmia. On admission to our hospital, vital signs and physical examination results were nonspecific. Serum lactic acid, creatine kinase-MB, troponin T, and N-terminal probrain natriuretic peptide were elevated to 32.2 mg/dL, 11.37 ng/mL, 2.287 ng/mL, and 2404 pg/mL, respectively. A transthoracic echocardiogram demonstrated severe concentric left ventricular hypertrophy (septal wall thickness of 15 mm, left ventricle posterior wall thickness of 15 mm, relative wall thickness of 0.55, and left ventricular mass index of 261.9 g/m²) with a diffusely speckled pattern of the myocardium (Figure 1A and 1B). Global hypokinesia of the left ventricle was evident, with an ejection fraction of 36%. Right ventricular hypertrophy and decreased systolic function were also noted (right ventricle free wall thickness, 8 mm; tricuspid annular plane systolic excursion, 14 mm). Coronary angiography showed normal coronary arteries. Cardiac MRI demonstrated diffuse biventricular hypertrophy and decreased left ventricle systolic function. T2-weighted imaging revealed focal increased signal in the midventricle and apex of the left ventricle. Late gadolinium enhancement imaging revealed focal increased signal in the midventricle and apex of the left ventricle systolic function. T2-weighted imaging revealed focal increased signal in the midventricle and apex of the left ventricle. Late gadolinium enhancement imaging revealed focal increased signal in the midventricle and apex of the left ventricle. Late gadolinium enhancement imaging revealed focal increased signal in the midventricle and apex of the left ventricle.

Images in Cardiovascular Medicine

Hypertrophic Cardiomyopathy Attributable to Mitochondrial DNA Mutation Diagnosed by Pathology and Gene Sequencing

Young Choi, MD; Jeong-Hwa Lee, MD, PhD; Mei Nu Cui, MD; Young Su Lee, MD; Mi-Hyang Jung, MD; Jeong-Eun Yi, MD; Hae Ok Jung, MD, PhD; Ho-Joong Youn, MD, PhD

Hypertrophic cardiomyopathy (HCM) is a genetic disorder characterized by myocardial hypertrophy and usually systolic dysfunction. It can be caused by mutations in sarcomeric genes, mitochondrial DNA (mtDNA) mutations, or non-sarcomeric genes. In this case, the diagnosis of mitochondrial cardiomyopathy was made after the identification of a mtDNA mutation in the patient. The mtDNA mutation identified was m.3243A>G, a mutation that has been previously associated with mitochondrial cardiomyopathy. The patient's symptoms included exertional dyspnea and arrhythmia, and his clinical presentation was consistent with a diagnosis of hypertrophic cardiomyopathy. The genetic testing, including mtDNA sequencing, confirmed the diagnosis of mitochondrial cardiomyopathy. The patient was treated with antihypertensive medications, including an angiotensin II receptor blocker, and his symptoms improved. This case demonstrates the importance of considering mitochondrial disease in the differential diagnosis of hypertrophic cardiomyopathy.
Disclosures

None.

References


Figure 1. Transthoracic echocardiogram and cardiac MRI. Two-dimensional echocardiogram in parasternal long axis (A) and parasternal short axis (B) showed concentric LV hypertrophy with a diffusely speckled pattern of the myocardium. Late gadolinium enhancement cardiac MRI revealed multifocal and curvilinear patchy enhancement in the subepicardial and midmyocardial layers on the long axis (C) and short axis (D), denoted by the white arrows. LV indicates left ventricle.

Figure 2. Endomyocardial biopsy findings. A, Light microscopy demonstrated cardiomyocyte hypertrophy and perinuclear vacuolization (black arrows; hematoxylin and eosin, ×40 original magnification). B, Electron microscopy revealed proliferation of polymorphic mitochondria causing displacement of myofibrils (×10,000). C, Electron microscopy images of variable-sized swollen mitochondria with abnormal cristae (×20,000).
Figure 3. Sequencing chromatogram of mitochondrial DNA mutation of leukocytes. **Top,** Shows the wild-type nucleotide G at the position 3243 in the *MT-TL1* gene. **Bottom,** Shows the heteroplasmic mutant allele A.
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