LETTER TO THE EDITOR

Left ventricular hypertrophy: A rare cardiac involvement of Becker muscular dystrophy

Dear Editor,

Due to a different mutation site in the Duchenne muscular dystrophy (DMD) gene, patients with Becker muscular dystrophy (BMD) show a milder and slower clinical progression as compared with patients with DMD. Cardiac involvement is a frequent feature of both dystrophies, which can develop before or after skeletal muscle damage. Asymptomatic cardiomyopathy is seen in most cases, and left- or bi-ventricular dilation occurs in up to one-third of patients with BMD [1]. In contrast, left ventricular (LV) hypertrophy has rarely been reported.

A 32-year-old male patient was admitted with the chief complaint of exertional dyspnea and fatigue accompanied by mild muscular pain and weakness in the lower extremities for 2 years. Mildly enlarged cardiac dullness, pseudo-hypertrophy of the calf muscles, Gowers sign, and mild muscular weakness and tenderness in the lower extremities were present on physical examination.

Laboratory tests showed very high levels of myogenic and myocardiogenic enzymes, including a cytokinase (CK) of 4730 IU/L (normal range: 19–226 IU/L), lactate dehydrogenase of 838 IU/L (110–220 IU/L), myoglobin of 966 ng/mL (28–72 ng/mL), CK-MB > 300 ng/L (< 4.94 ng/L), and Troponin-T of 80 ng/L (< 14 ng/L). The level of serum N-terminal-pro-brain natriuretic peptide was 512 pg/mL (0–88 pg/mL). The echocardiogram (ECG) demonstrated LV hypertrophy with increased R-wave amplitude and inverted T-wave in the left-sided ECG leads (V5, V6, I, and AVL). Two-dimensional echocardiography revealed severe LV hypertrophy, normal LV end-diastolic dimensions, and a preserved LV-ejection fraction. Neither an outflow tract obstruction nor mitral leaflet systolic anterior motion was detected (Figure 1A). Moreover, delayed enhancement of the myocardium was detected by cardiac magnetic resonance (CMR) (Figure 1B). The electromyogram displayed myogenic damage, but normal nerve conduction. Consequently, a skeletal muscle biopsy with specific immunohistochemistry showed positive expression of dysferlin and α, β, γ, and δ-sarcoglycan, but the intensity indicated decreased N-terminal, C-terminal, and rod-domain dystrophin, which helped to confirm BMD (Figure 1C as compared to normal skeletal muscle cells from another patient shown in Figure 1D; informed consent was obtained from the patients to publish their data). Unfortunately, this patient refused genetic testing.

X-linked DMD and BMD are allelic disorders caused by mutations in the DMD gene. The gene mainly encodes dystrophin, which is primarily localized at the surface membrane of striated muscle fibers and is expressed in similar isoforms in skeletal, cardiac, and smooth muscle. In DMD, dystrophin is nearly absent. Severe skeletal muscle involvement occurs at a very young age and rapidly progresses to akinesia within a few years. In contrast, dystrophin remains present at some concentration, but shows an abnormal phenotype in BMD [1]. Similar to this case, BMD patients usually present later in life, with mild skeletal muscular symptoms of slower progression; however, these patients usually have obvious symptoms of cardiac involvement.

Because a dystrophin deficiency makes muscular cells vulnerable to the damage caused by mechanical stress, necrotic myocytes are replaced by connective tissue fibers and fat [2]. However, there is great variability in the degree of cardiac involvement in BMD. As previously reported, different types of ECG abnormalities are frequently observed in BMD. Not surprisingly, morphological changes in the myocardium usually manifest as LV or biventricular chamber dilation [3]. Delayed gadolinium enhancement in the dilated LV via CMR was also reported [4]. In contrast, LV hypertrophy (as in this case) is extremely rare in both BMD and DMD. There was one case reported of a 17-year-old boy with BMD who presented with a rapid progression from hypertrophic cardiomyopathy to heart failure within 2 years [5]. Therefore, myopathy should be considered when LV...
hypertrophy is accompanied by myalgia, difficulty in moving, and increased myogenic enzymes. Muscle biopsy and genetic testing may help confirm the diagnosis.

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References


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