Electron microscopic findings of cardiomyopathy with limb girdle muscular dystrophy

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Abstract

Cardiac involvement in limb girdle muscular dystrophy has considered to be rare. This is the first report showing the electron microscopic findings of dilated cardiomyopathy (DCM) accompanied with limb girdle muscular dystrophy. The findings described in this report indicate that limb girdle muscular dystrophy may be yet another cause of DCM.

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1. Case 1

A 53-year-old male was referred for the evaluation of cardiac dysfunction. He was diagnosed as limb girdle muscular dystrophy on muscle biopsy 15 years ago. There was no history of muscular dystrophy in the family. The pulse was 90 beats per minute and the blood pressure was 100/70 mm Hg. No cardiac murmur was heard and there was slight peripheral edema. An electrocardiography showed ectopic atrial rhythm and multifocal premature ventricular contractions. An echocardiography disclosed a diffuse hypokinesis with ejection fraction 0.4 and dilated left ventricle with thin wall. The coronary angiography revealed normal coronary arteries. Electron microscopic findings of the specimen obtained by left ventricular endomyocardial biopsy demonstrated a disarray of extremely stretched myocytes without inflammatory cells (Fig. 1A). The mitochondria showed decreases in size with pleomorphism, and alterations in the density of the matrix (Fig. 1B). Pathological findings supported the diagnoses of dilated cardiomyopathy (DCM).

2. Case 2

A 58-year-old female who has been given a diagnosis of limb girdle muscular dystrophy since age 27 admitted to our hospital due to congestive heart failure. There was no history of consanguinity or neuromuscular disease in the family. The blood pressure was 130/80 mm Hg and pansystolic murmur was heard at apex. An electrocardiography showed ectopic atrial rhythm, abnormal Q in aVL, and ST depression in V6. Echocardiography revealed a diffuse hypokinesis with ejection fraction 0.3 and dilated left ventricle with thin wall. The coronary angiography revealed normal coronary arteries. Electron microscopic findings of the specimen obtained by left ventricular endomyocardial biopsy showed myocyte hypertrophy, mitochondrial pleomorphism, and nuclei with highly lobulated membranes, which displaced adjacent myofibrils and caused breakdown of normal Z-band registration (Fig. 2). Myofibrils coursed in several different directions without inflammatory cells. These pathological findings also supported the diagnoses of DCM.

3. Discussion

Cardiac involvement in limb girdle muscular dystrophy has considered to be rare [1–5]. As more has been
understood about the genetic constitution and heterogeneity of this group of disorders, a greater realization of the potential for cardiac involvement is emerging. Cardiac manifestations include conduction system disease and arrhythmias; sudden cardiac death is a recognized complication, and pacemaker placement is often necessary. However, DCM is extremely uncommon in patients with limb girdle muscular dystrophy[2,4,5]. Recently, it has been reported using echocardiography that 3 of 42 patients with merosin-positive congenital muscular dystrophy related to the autosomal recessive limb girdle muscular dystrophy has ejection fraction between 50% and 55%, suggesting the existence of left ventricular dysfunction[6]. We demonstrated in the present cases that the degree of left ventricular systolic function was more severe than those reports and the histological findings of the heart were compatible with dilated cardiomyopathy. This is the first report showing the electron microscopic findings of DCM accompanied with limb girdle muscular dystrophy. The findings described in this report indicate that limb girdle muscular dystrophy may be yet another cause of DCM.

References


Fig. 1. (A, B) Electron microscopic findings of the specimen obtained by left ventricular endomyocardial biopsy in “Case 1”.

Fig. 2. Electron microscopic findings of the specimen obtained by left ventricular endomyocardial biopsy in “Case 2”.