Abstract

BACKGROUND: Dystrophin-glycoprotein complex (DGC)-related muscular dystrophies may present similar clinical and pathological features as well as undetectable mutations thus being sometimes difficult to distinguish. We investigated the value of muscle magnetic resonance imaging (MRI) in the differential diagnosis of DGC-related muscular dystrophies and reported the largest series of Chinese patients with sarcoglycanopathies studied by muscle MRI.

RESULTS: Fifty-five patients with DGC-related muscular dystrophies, including 22 with confirmed sarcoglycanopathies, 11 with limb-girdle muscular dystrophy 2I (LGMD2I, FKRP-associated dystroglycanopathy), and 22 with dystrophinopathies underwent extensive clinical evaluation, muscle biopsies, genetic analysis, and muscle MRI examinations. Hierarchical clustering of patients according to the clinical characteristics showed that patients did not cluster according to the genotypes. No statistically significant differences were observed between sarcoglycanopathies and LGMD2I in terms of thigh muscle involvement. The concentric fatty infiltration pattern was observed not only in different sarcoglycanopathies (14/22) but also in LGMD2I (9/11). The trefoil with single fruit sign was observed in most patients with dystrophinopathies (21/22), and a few patients with
sarcoglycanopathies (4/22) or LGMD2I (2/11). Hierarchical clustering showed that most patients with sarcoglycanopathies or LGMD2I can be distinguished from dystrophinopathies based on the concentric fatty infiltration pattern and trefoil with single fruit sign at the thigh level on muscle MRI.

**CONCLUSIONS:** Muscle MRI at the thigh level potentially allows distinction of sarcoglycanopathies or FKRP-associated dystroglycanopathy from dystrophinopathies.

**KEYWORDS:** Differential diagnosis; Dystrophin-glycoprotein complex; Muscle magnetic resonance imaging; Muscular dystrophy

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