Limb-girdle muscular dystrophy type 2I: two Chinese families and a review in Asian patients.

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Abstract

BACKGROUND: Limb-girdle muscular dystrophy type 2I (LGMD2I) is an autosomal recessive hereditary disorder caused by mutations in the fukutin-related protein (FKRP) gene. Although the features of the disorder in European patients have been summarized, Asian patients with LGMD2I have rarely been reported. Thus, the clinical differences in LGMD2I between Asian and European patients and the associated genetic changes remain unclear.

METHODS: We reported detailed clinical data as well as results from muscle biopsy, muscle MRI and genetic analysis of the FKRP gene in two unrelated Chinese families with LGMD2I. Additionally, a review of the literature focusing on the clinical and mutational features of LGMD2I in Asian patients was performed.

RESULTS: The muscle biopsy results showed dystrophic features. Immunohistochemical staining revealed decreased glycosylations on α-dystroglycan. The muscle MRI results showed that the gluteus maximus, adductor, biceps femoris, vastus intermedius and vastus lateralis were severely affected. The patients in the two families harbored the same compound heterozygous mutations (c.545A>G and c.948delC). One patient showed significant clinical improvement after corticosteroid treatment.

CONCLUSION: Our study expanded the reported spectrum of Asian LGMD2I patients. Our literature review revealed that pathogenic mutations in the FKRP gene in Asian LGMD2I patients are compound heterozygous rather than homozygous. Compound heterozygous Asian patients have a mild phenotype but frequently show respiratory and cardiac impairments. Corticosteroids may be beneficial for the treatment of LGMD2I and should be further investigated.

KEYWORDS: FKRP gene; Limb-girdle muscular dystrophy type 2I; MRI; clinical phenotype; steroid treatment; α-dystroglycan