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Genetic Variations and Clinical Spectrum of Dystroglycanopathy in a Large Cohort of Chinese Patients

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Abstract

Dystroglycanopathy is a group of muscular dystrophies with deficient glycosylation of alpha-dystroglycan (α-DG). We recruited patients from 36 tertiary academic hospitals in China. In total, 143 patients with genetically diagnosed dystroglycanopathy were enrolled. Of these, limb girdle muscular dystrophy was the most common initial diagnosis (83 patients) and Walker-Warburg syndrome was the least common (1 patient). In 143 patients, mutations in FKRP gene were the most prevalent (62 patients), followed by POMT2 (16), POMT1 (16), POMGNT1 (14), ISPD (14), FKTN (9), GMPPB (7), B3GALNT2 (3), DPM3 (1) and DAG1 (1). Several frequent mutations were identified in FKRP, POMT1, POMGNT1, ISPD, and FKTN genes. Many of these were founder mutations. Patients with FKRP mutations tended to have milder phenotypes, while those with mutations in POMGNT1 genes had more severe phenotypes. Mental retardation was a clinical feature associated with mutations of POMT1 gene. Detailed clinical data of 83 patients followed up in Peking University First Hospital were further analyzed. Our clinical and genetic analysis of a large cohort of Chinese patients with dystroglycanopathy expanded the genotype variation and clinical spectrum of congenital muscular dystrophies.

Keywords: dystroglycanopathy; genotype constituent ratio; phenotype; population study.

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