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J Hum Genet. 2016 Aug;61(8):753-9. doi: 10.1038/jhg.2016.42. Epub 2016 May 19.

Analysis of phenotype, enzyme activity and genotype of Chinese patients with POMT1 mutation.

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Abstract

Protein O-mannosyltransferase 1 (POMT1) is a glycosyltransferase involved in α -dystroglycan glycosylation. POMT1 mutations cause a wide spectrum of clinical conditions from Walker-Warburg syndrome (WWS), which involves muscle, eye and brain abnormalities, to mild forms of limb-girdle muscular dystrophy with mental retardation. We aimed to elucidate the impact of different POMT1 mutations on the clinical phenotype. We report five Chinese patients with POMT1 mutations: one had a typical clinical manifestation of WWS, and the other four were diagnosed with congenital muscular dystrophy with mental retardation of varying severity. We analyzed the influence of the POMT1 mutations on POMT activity by assaying the patients' muscles and cultured skin fibroblasts. We demonstrated different levels of decreased POMT activity that correlated highly with decreased α -dystroglycan glycosylation. Our results suggest that POMT activity is inversely proportional to clinical severity, and demonstrate that skin fibroblasts can be used for differential diagnosis of patients with α -dystroglycanopathies. We have provided clinical, histological, enzymatic and genetic evidence of POMT1 involvement in five unrelated Chinese patients.

PMID: 27193224 DOI: [10.1038/jhg.2016.42](https://doi.org/10.1038/jhg.2016.42)

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