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Clinical and genetic diversity of nemaline myopathy from a single neuromuscular center in Korea.

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Abstract

Nemaline myopathy (NM), the most common of the congenital myopathies, is caused by various genetic mutations. In this study, we attempted to identify the causative mutations of NM and to reveal any specific genotype-phenotype relationship in Korean patients with this disease. We investigated the clinical features and genotypes in 15 pathologically diagnosed NM patients, using whole exome sequencing (WES) combined with targeted sequencing and array-based comparative genomic hybridization. This strategy revealed pathogenic causative mutations in seven patients (46.7%), among whom mutations in the nebulin gene (NEB) were the most frequent (5 patients, 33.3%). Copy number variation (CNV) abnormality in NEB was not observed in any of our patients. In those with NEB-associated NM, the clinical spectrum was highly variable regardless of the mutation type. However, the majority of patients showing anterior lower leg weakness were associated with mutations located between NEB exons 166 and 177. We concluded that the combination of WES and targeted Sanger sequencing is an effective strategy for analyzing genotypes in patients with NM, and that CNV in NEB may not be a frequent cause of this disease among Koreans.

KEYWORDS: Comparative genomic hybridization; Copy number variation; NEB; Nemaline myopathy; Whole exome sequencing

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