
**Format:** Abstract


**Mutational and clinical spectrum in a cohort of Chinese patients with hereditary nemaline myopathy.**

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**Abstract**

Hereditary nemaline myopathy (NM) is one of the most common congenital myopathies with the histopathological findings of nemaline bodies. We used targeted next generation sequencing (NGS) to identify causative mutations in 48 NM patients with confirmed myopathological diagnosis, analyze the mutational spectrum and phenotypic features. Furthermore, reverse transcription PCR (RT-PCR) was used to confirm the pathogenic effect of one NEB splicing variant. The results showed that variants were found in five NM-associated genes, including NEB, ACTA1, TNNT1, KBTBD13, and CFL2, in 34 (73.9%), 7 (15.2%), 3 (6.5%), 1 (2.2%), and 1 (2.2%) patients, respectively, in a total of 46/48 (95.8%) NM patients. Of the total 64 variants identified, 51 were novel variants including 26 pathogenic, 1 likely pathogenic, and 24 with undetermined significance (VUS). Notably, one NEB splicing mutation, c.21417 + 3A > G causing exon 144 splicing (NM_001164508.1), as confirmed by RT-PCR, was found in 52.9% (18 patients) of NEB variant-carrying patients. Typical congenital NM, the most common clinical subtype (60.4%), was associated with 5 NM genes. We concluded that hereditary NM showed a highly variable genetic spectrum. NEB was the most frequent causative gene in this Chinese cohort, followed by ACTA1. We found a hotspot splicing mutation in NEB among Chinese cohort. This article is protected by copyright. All rights reserved.

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**KEYWORDS:** Chinese; Hereditary nemaline myopathy; genotype; phenotype
