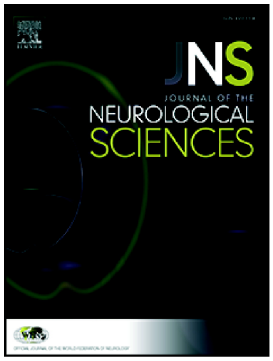

A patient with slowly progressive adult-onset nemaline myopathy and novel compound heterozygous mutations in the nebulin gene

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A patient with slowly progressive adult-onset nemaline myopathy and novel compound heterozygous mutations in the nebulin gene

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Highlights

- This case of adult-onset nemaline myopathy showed hypercapnic respiratory failure.
- Muscle weakness progressed very slowly in our patient.
- Novel compound heterozygous mutations identified in the NEB gene.

Key words: adult-onset nemaline myopathy, compound heterozygote, nebulin (NEB)

Abbreviations: ACTA1, alfa-actin; ALT, alanine aminotransferase; AS-PCR, allele specific-polymerase chain reaction; AST, aspartate aminotransferase; CFL2, cofilin2; CO_2, carbon dioxide; CT, computed tomography; DNA, deoxyribonucleic acid; ESP6500, Exome Sequencing Project 6500; H&E, hematoxylin and eosin; HCO_3^-, bicarbonate ion; HGVD, Human Genetic Variation Database; MGUS, monoclonal gammopathy of undetermined significance; NADH-TR, nicotinamide adenine dinucleotide tetrazolium reductase; NEB, nebulin; O_2, oxygen; PCO_2, carbon dioxide partial pressure; PCR-RFLP, polymerase chain reaction-restriction fragment length polymorphism; PO_2, oxygen partial pressure; SLONM, sporadic late onset nemaline myopathy; SO_2, oxygen saturation; TNNT1, troponin T1; TPM2, beta-tropomyosin; TPM3, alfa-tropomyosin; Xp, X-ray photography.