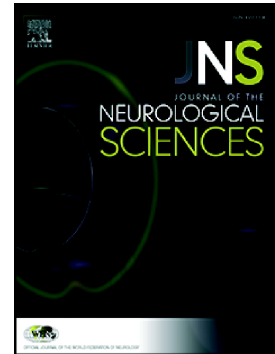


## Accepted Manuscript

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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<sub>2</sub>, carbon dioxide; CT, computed tomography; DNA, deoxyribonucleic acid; ESP6500, Exome Sequencing Project 6500; H&E, hematoxylin and eosin; HCO<sub>3</sub><sup>-</sup>, bicarbonate ion; HGVD, Human Genetic Variation Database; MGUS, monoclonal gammopathy of undetermined significance; NADH-TR, nicotinamide adenine dinucleotide tetrazolium reductase; NEB, nebulin; O<sub>2</sub>, oxygen; PCO<sub>2</sub>, carbon dioxide partial pressure; PCR-RFLP, polymerase chain reaction-restriction fragment length polymorphism; PO<sub>2</sub>, oxygen partial pressure; SLONM, sporadic late onset nemaline myopathy; SO<sub>2</sub>, oxygen saturation; TNNT1, troponin T1; TPM2, beta-tropomyosin; TPM3, alfa-tropomyosin; Xp, X-ray photography.

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