Supplementary Fig. 1. Histopathological findings in a biopsy of the deltoid muscle of the proband. The muscle fibers did not show significant necrosis or regenerating fibers, with well-organized intramyofibrillar networks (A and B). However, typical grouped fiber atrophy was observed, reflecting neurogenic changes (C, D, E). The dystrophin stain was normal (F) (A: Hematoxylin-eosin, ×200; B: NADH-tetrazolium reductase, ×100; C: Cytochrome c oxidase, ×100; D: ATPase 4.4, ×40; E: ATPase 4.4, ×100; and F: Dystrophin, ×200).