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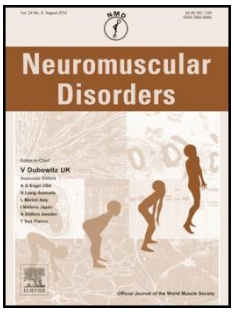

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Tubular aggregates in congenital myasthenic syndrome

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A 20-year-old man with history of fluctuating muscle weakness since early childhood was referred to our department. There was no history of ptosis, double vision or dysarthria and no cognitive impairment. A muscle biopsy at the age of 8 years was inconclusive. Electromyography was performed at the age of 7 and 12 years with evidence of myopathy in proximal muscles. Neuromuscular transmission was not tested at that time. A muscle biopsy at age 17 showed myopathic changes, tubular aggregates and autophagic vacuoles. The tubular aggregates demonstrated