Non-compaction cardiomyopathy and early respiratory failure in an adult symptomatic female carrier of centronuclear myopathy caused by a MTM1 mutation.


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Abstract
X-linked myotubular myopathy (XLMTM) is a rare neuromuscular condition caused by mutations in the MTM1 gene. Female carriers are believed to be usually asymptomatic; nevertheless, recent reports have displayed a wide a spectrum of clinical involvement in females suggesting that MTM1 mutations might be underestimated in this population. Here we report a 55-year-old woman manifesting with an abrupt respiratory decline, whose respiratory function tests revealed a severe restrictive ventilatory defect. The neurological examination identified mild proximal leg weakness and her cardiac evaluation showed a non-compaction cardiomyopathy with normal left ventricle function. Muscle biopsy was consistent with centronuclear myopathy. Next-generation sequencing of 49 genes related to congenital myopathies allowed the identification of a 4bp deletion in the MTM1 gene, leading to a truncating mutation previously described in males but for the first time reported in a female patient.

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