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## Whole exome sequencing discloses a pathogenic MTM1 gene mutation and ends the diagnostic odyssey in an older woman with a progressive and seemingly sporadic myopathy: Case report and literature review of MTM1 manifesting female carriers.

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### Abstract

We report the case of a 58-year-old woman with a progressive and seemingly sporadic myopathy who, later through whole exome sequencing, was diagnosed as a manifesting carrier of a myotubularin 1 gene mutation (c.342\_342 + 4delAGTAA). As the case was a diagnostic challenge for 7 years, we thought it would be helpful to report the patient and review the other 25 cases thus far described. The manifesting carrier state is a rare cause for myopathic weakness in a female but should be strongly considered in kindreds with known affected males with myotubularin 1 gene mutations, and families with history of gestational polyhydramnios or male infantile death. Although the clinical phenotype is quite variable, the findings of ptosis, ophthalmoparesis, facial weakness, and asymmetrical limb involvement should raise the suspicion of the manifesting carrier state. Necklace fibers appear to be a highly sensitive and specific pathologic finding in such cases.

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