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# Myopathy due to Myoadenylate Deaminase Deficiency

National Cancer Institute

## Source

National Cancer Institute. *Myopathy due to Myoadenylate Deaminase Deficiency*. NCI Thesaurus. Code C157504.

An autosomal recessive condition caused by mutation(s) in the AMPD1 gene, encoding AMP deaminase 1. The condition is characterized by exercise-induced muscle pain and/or fatigue, which may be associated with rhabdomyolysis and/or increased concentrations of creatinine kinase.