

Autosomal Dominant Distal Myopathy With Nemaline Rods Due to p.Glu197Asp Mutation in ACTA1

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PMID: 30732915 DOI: 10.1016/j.nmd.2018.12.001

Abstract
In a previous report of a new phenotype with predominant scapulo-humeral-peroneal-distal myopathy associated with the Glu197Asp mutation in ACTA1, muscle biopsies did not show nemaline rods, nor could nemaline rods formation be demonstrated in an exhaustive functional in vivo or in vitro study. However, muscle biopsy in members of our family, carrying a similar clinical phenotype of some members of the original family and the same ACTA1 mutation, revealed the presence of numerous nemaline rods, suggesting that there must be other factors that explain the absence of nemaline rods.

Keywords: ACTA1; Congenital myopathy; Distal; Nemaline rod.

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