Recessive MYPN mutations cause cap myopathy with occasional nemaline rods.


Abstract

Congenital myopathies are phenotypically and genetically heterogeneous. We describe homozygous truncating mutations in MYPN in 2 unrelated families with a slowly progressive congenital cap myopathy. MYPN encodes the Z-line protein myopalladin implicated in sarcomere integrity. Functional experiments demonstrate that the mutations lead to mRNA defects and to a strong reduction in full-length protein expression. Myopalladin signals accumulate in the caps together with alpha-actinin. Dominant MYPN mutations were previously reported in cardiomyopathies. Our data uncover that mutations in MYPN cause either a cardiac or a congenital skeletal muscle disorder through different modes of inheritance. This article is protected by copyright. All rights reserved.

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