Myofibrillar myopathy caused by a novel FHL1 mutation presenting a mild myopathy with ankle contracture.

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Abstract

FHL1-related myopathies are clinically heterogeneous, involving skeletal and cardiac muscles. Overlapping clinical features include joint contractures, rigid spine, scapuloperoneal weakness and cardiac diseases. Histopathologically, reducing bodies are the most characteristic finding, but not present in all FHL1-related cases. Non-specific dystrophic pathology without reducing body is usual in the forms of X-linked myopathy with postural muscle atrophy, Emery-Dreifuss muscular dystrophy and isolated hypertrophic cardiomyopathy. Here, we describe a patient with mild weakness with ankle contracture. We finally concluded he has a FHL1-related myopathy at an extreme end of phenotypic spectrum of FHL1 myopathy, which one might miss to recognize as a form of myopathy. The genetic variant was detected by whole exome sequencing, and its pathogenicity was clearly confirmed with pathological and biochemical studies. This is the first FHL1 case with a mildest phenotype backed by biochemical/genetic evidence. This report will help clinicians hesitating to further evaluate mild cases to better correlate the genotype to the phenotype.

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