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Clinical and imaging hallmarks of the MYH7-related myopathy with severe axial involvement.

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Abstract

INTRODUCTION: MYH7 gene mutations are related to a heterogeneous group of skeletal and cardiac myopathies.

METHODS: We evaluated clinical and muscle magnetic resonance imaging changes in patients with mutations in the rod domain of MYH7 including one with mosaicism and three with novel missense mutations.

RESULTS: Patients presented in childhood with a distal and axial phenotype. Biopsy findings were variable. Half of the cases displaying some type of core pathology, including minicores and eccentric cores. Most patients demonstrated internal bands of infiltration ("inverted-collagen-VI sign") in multiple muscles, particularly the soleus, and prominent atrophy and fatty infiltration of the tongue and the paraspinal, gluteus minimus, sartorius, gracilis, tibialis anterior and extensor digitorum longus muscles.

DISCUSSION: Muscle imaging findings in patients with axial involvement provide significant clues permitting the distinction between MYH7-related myopathies and other axial myopathies such as those related to SEPN1 and LMNA genes. This article is protected by copyright. All rights reserved.

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KEYWORDS: LMNA; MYH7; SEPN1; foot drop; heatmap; muscle imaging; rigid spine

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