Importance of early diagnosis in LMNA-related muscular dystrophy for cardiac surveillance.

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Abstract

INTRODUCTION: The identification of LMNA-related muscular dystrophy is important because it poses life-threatening cardiac complications. However, diagnosis of LMNA-related muscular dystrophy based on clinical features is challenging.

METHODS: We reviewed the clinical phenotypes of 14 children with LMNA variants, focusing on the cardiac function and genotypes.

RESULTS: Most patients presented with motor developmental delay or gait abnormalities. Eight (57%) patients had prominent neck extensor weakness or contractures. All patients showed ankle contractures at an early stage. Regular cardiac surveillance allowed for the detection of dysrhythmias in 57% of patients at a mean age of 14 years (range, 5-26). All patients had missense variants; however, there were no clear phenotype-genotype correlations.

DISCUSSION: Early diagnosis of LMNA-related muscular dystrophy provides an opportunity for cardiac surveillance, potentially leading to the prevention of cardiac mortality in children.

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