The 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 the 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 patients (57%) had prominent neck extensor weakness or contractures. All patients showed ankle contractures in 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 years). 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...
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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