Dropped head congenital muscular dystrophy caused by de novo mutations in LMNA.


Abstract

BACKGROUND: Dropped head syndrome is an easily recognizable clinical presentation of Lamin A/C-related congenital muscular dystrophy. Patients usually present in the first year of life with profound neck muscle weakness, dropped head, and elevated serum creatine kinase.

CASE DESCRIPTION: Two patients exhibited head drop during infancy although they were able to sit independently. Later they developed progressive axial and limb-girdle weakness. Creatine kinase levels were elevated and muscle biopsies of both patients showed severe dystrophic changes. The distinctive clinical hallmark of the dropped head led us to the diagnosis of Lamin A/C-related congenital muscular dystrophy, with a pathogenic de novo mutation p.Glu31del in the head domain of the Lamin A/C gene in both patients. Remarkably, one patient also had a central involvement with white matter changes on brain magnetic resonance imaging.

CONCLUSION: Lamin A/C-related dropped-head syndrome is a rapidly progressive congenital muscular dystrophy and may lead to loss of ambulation, respiratory insufficiency, and cardiac complications. Thus, the genetic diagnosis of dropped-head syndrome as L-CMD and the implicated clinical care protocols are of vital importance for these patients. This disease may be underdiagnosed, as only a few genetically confirmed cases have been reported.

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KEYWORDS: Dropped head; LMNA-related congenital muscular dystrophy; Lamin A/C gene; White matter

PMID: 27876398 DOI: 10.1016/j.braindev.2016.11.002

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