
Turner C¹, Mein R², Sharpe C³, Love DR⁴.

Abstract

Merosin deficient congenital muscular dystrophy (MDC1A) is an autosomal recessive disorder characterized by mutations in the LAMA2 gene at chromosome 6q22-23. This gene spans 65 exons and encodes the α2 chain subunit of laminin-2. A variety of deletions, missense, nonsense and splice site mutations have been described in the LAMA2 gene, with resultant MDC1A. We describe a novel LAMA2 homozygous sequence variant in a Samoan patient with MDC1A and confirm its pathogenic effect with merosin immunohistochemistry on skeletal muscle biopsy. The likely effect of the sequence variant is modeled using in silico analysis.

KEYWORDS: Immunohistochemistry; Laminin-2; Merosin deficient congenital muscular dystrophy; Mutation

PMID: 26249246 DOI: 10.1016/j.jocn.2015.04.016

[Indexed for MEDLINE]