Genotype-phenotype correlation in a large population of muscular dystrophy patients with LAMA2 mutations.


Abstract
Merosin deficient congenital muscular dystrophy 1A (MDC1A) results from mutations in the LAMA2 gene. We report 51 patients with MDC1A and examine the relationship between degree of merosin expression, genotype and clinical features. Thirty-three patients had absence of merosin and 13 showed some residual merosin. Compared to the residual merosin group, patients with absent merosin had an earlier presentation (<7days) (P=0.0073), were more likely to lack independent ambulation (P=0.0215), or require enteral feeding (P=0.0099) and ventilatory support (P=0.0354). We identified 33 novel LAMA2 mutations; these were distributed throughout the gene in patients with absent merosin, with minor clusters in exon 27, 14, 25 and 26 (55% of mutations). Patients with residual merosin often carried at least one splice site mutation and less frequently frameshift mutations. This large study identified novel LAMA2 mutations and highlights the role of immunohistochemical studies for merosin status in predicting clinical severity of MDC1A.

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