COL6A and LAMA2 Mutation Congenital Muscular Dystrophy: A Clinical and Electrophysiological Study.

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Abstract

OBJECTIVES: COL6A and LAMA2 are subtypes of congenital muscular dystrophy.

METHODS: Retrospective chart review of clinical findings, spirometry, muscle histology, muscle ultrasound, neuroimaging, and Electromyography (EMG)/Nerve Conduction Study data in genetically confirmed COL6A and LAMA2 subjects.

RESULTS: We identified 8 COL6A and 6 LAMA2 subjects: the female-to-male ratio was 1.3:1 and the mean age was 11.9 ± 3.6 years. Gross motor delays since birth, proximal muscle weakness, and contractures were noted in both groups. Joint hyperlaxity and skin changes (follicular hyperkeratosis and muscle biopsy scar thinning) were unique to COL6A. Severe scoliosis, macrocephaly, and nonambulatory status were common in LAMA2. Increasing age was associated with poor respiratory function in COL6A. There was central "cloud appearance" on rectus femoris muscle ultrasound in COL6A and white matter T2 hyperintensity on brain magnetic resonance imaging in LAMA2. LAMA2 also showed demyelinating polyneuropathy. Neurogenic changes on EMG and muscle histology were noted in 37% and 33% of COL6A cases, respectively.

CONCLUSIONS: COL6A has unique skin changes, central cloud appearance on muscle ultrasound. LAMA2 has demyelinating polyneuropathy and white matter changes on brain imaging. The presence of neurogenic changes on EMG and muscle histology in COL6A suggests motor axonal neuropathy. Genetic testing remains the gold standard in confirming COL6A congenital muscular dystrophy.

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