Comparison of sitting and supine forced vital capacity in collagen VI-related dystrophy and laminin α2-related dystrophy.

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Abstract

BACKGROUND: Progressive, restrictive, respiratory insufficiency is the major cause of morbidity and mortality in Congenital Muscular Dystrophy (CMD). Nocturnal hypoventilation precedes daytime alveolar hypoventilation, and if untreated, may lead to respiratory failure and cor pulmonale. CMD consensus care guidelines recommend screening for respiratory insufficiency by conventional and dynamic (sitting to supine) pulmonary function testing (PFT) and evaluating for sleep disordered breathing if there is more than 20% relative reduction from sitting to supine FVC(L) (ΔFVC).

OBJECTIVE: The objective of this retrospective study was to explore and characterize dynamic FVC measures in 51 individuals with two common subtypes of CMD, COL6-RD, and LAMA2-RD.

METHODS: We compared sitting and supine FVC in patients with confirmed mutation(s) in either COL6 or LAMA2. We investigated influences of age, CMD subtype, gender, race, ambulatory status, and non-invasive positive pressure ventilation (NIPPV) status on FVC percent predicted (FVCpp) and ΔFVC.

RESULTS: COL6-RD participants exhibited a significant difference between sitting and supine mean FVCpp (sitting 66.1, supine 55.1; P < 0.0001) and were 5.4 times more likely to have -ΔFVC >20% than those with LAMA2-RD when controlling for ambulant status. FVCpp sitting correlated inversely with age in individuals ≤18 years.

CONCLUSION: FVCpp sitting decreases progressively in childhood in both CMD subtypes. However, our results point to a difference in diaphragmatic involvement, with COL6-RD individuals having more disproportionate diaphragmatic weakness than LAMA2-RD. A ΔFVC of greater than -20% should continue to be used to prompt evaluation of sleep-disordered breathing. Timely initiation of NIPPV may be indicated to treat nocturnal hypoventilation. Pediatr Pulmonol. © 2017 Wiley Periodicals, Inc.
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KEYWORDS: congenital muscular dystrophy; dynamic spirometry; neuromuscular disorders; pulmonary function testing; vital capacity

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