Diaphragmatic dysfunction in Collagen VI myopathies.

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Abstract

Collagen VI-related myopathies are hereditary disorders causing progressive restrictive respiratory insufficiency. Specific diaphragm involvement has been suggested by a drop in supine volumes. This pilot study aimed at characterizing the respiratory muscle phenotype in patients with COL6A1-3 genes mutations. Lung function, blood gases, muscle strength and respiratory mechanics were measured in 7 patients between 2002 and 2012. Patients were classified as Early-Severe (n = 3), Moderate-Progressive (n = 2) and Mild (n = 2) according to clinical disease presentation. Seven patients (aged 6-28) were evaluated. Forced vital capacity distinguished the Mild group (>60\% predicted) from the two other groups (<50\% predicted). This distinction was also possible using the motor function measure scale. Diaphragmatic dysfunction at rest was observed in all the Early-Severe and Moderate-Progressive patients. During a voluntary sniff maneuver diaphragmatic dysfunction was observed in all patients, as assessed by a negative gastric pressure. All patients had diaphragmatic fatigue assessed by a tension-time index over the threshold of 0.15. Diaphragmatic dysfunction during a maximal voluntary maneuver and diaphragmatic fatigue are constant features in Collagen VI myopathies. These observations can assist the diagnosis and should be taken in account for the clinical management, with the early detection of sleep-disordered breathing.

KEYWORDS: Bethlem myopathy; COLVI-related myopathy; Diaphragm; Motor function measure scale; Respiratory function; Ullrich \textit{congenital muscular dystrophy}; Whole body muscle magnetic resonance imaging

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