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Title: Diaphragmatic dysfunction in collagen VI myopathies

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Body: Introduction Collagen VI (COLVI)-related myopathies are hereditary disorders causing progressive restrictive respiratory insufficiency. Specific diaphragm involvement has been suggested by a drop in supine volumes. Objectives This pilot study aimed at characterizing the clinical respiratory muscle phenotype in patients with COL6A1-3 genes mutations. Methods Lung function, blood gases, muscle strength and respiratory mechanics were measured in patients with genetically confirmed COLVI myopathy between 2002 and 2012. Patients were classified as Early Severe, Moderate Progressive and Mild according to clinical disease presentation. Results Seven patients with mutations in the COL6A genes of variable severity (2 Mild, 2 Moderate Progressive and 3 Early Severe) were evaluated at a mean age of 12.0±7.7 years. Forced vital capacity distinguished the Mild group (>60% predicted) from the 2 other groups (<50% predicted). This distinction of severity was also possible using the motor function measure scale. The most striking observation was a diaphragmatic dysfunction in all patients during a voluntary maneuver, as assessed by a negative gastric pressure (Pgas) during a maximal sniff maneuver. Diaphragmatic dysfunction at rest was observed only in the Early Severe and Moderate Progressive patients with a negative Pgas during inspiration. All patients had diaphragmatic fatigue assessed by a diaphragmatic

tension-time index over the threshold of 0.15. Conclusion Diaphragmatic dysfunction during maximal voluntary maneuver and diaphragmatic fatigue are constant features in COLVI 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.