Hereditary Myopathies with Early Respiratory Insufficiency in Adults.

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Abstract

INTRODUCTION: Hereditary myopathies with early respiratory insufficiency as a predominant feature of the clinical phenotype are uncommon and underestimated in adults.

METHODS: We reviewed the clinical and laboratory data of patients with hereditary myopathies that demonstrated early respiratory insufficiency prior to the need for ambulatory assistance. Only patients with disease-causing mutations or a specific histopathological diagnosis were included. Patients with cardiomyopathy were excluded.

RESULTS: We identified 22 patients; half had isolated respiratory symptoms at onset. The diagnosis of the myopathy was often delayed resulting in delayed ventilatory support. The most common myopathies were adult-onset Pompe disease, myofibrillar myopathy, multiminicore disease and myotonic dystrophy-type 1. Single cases of laminopathy, MELAS, centronuclear myopathy and cytoplasmic body myopathy were identified.

DISCUSSION: We highlighted the most common hereditary myopathies associated with early respiratory insufficiency as the predominant clinical feature, and underscored the importance of a timely diagnosis for patient care. This article is protected by copyright. All rights reserved.

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KEYWORDS: congenital myopathy; diaphragm weakness; early respiratory insufficiency; hereditary myopathy; neuromuscular respiratory failure

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