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 who demonstrated early respiratory insufficiency before 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, multi-minicore disease, and myotonic dystrophy type 1. Single cases of laminopathy, MELAS (mitochondrial encephalomyopathy with lactic acidosis and strokelike events), centronuclear myopathy, and cytoplasmic body myopathy were identified.

CONCLUSION: 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. Muscle Nerve 56: 881-886, 2017.

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

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