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## Muscular MRI-based algorithm to differentiate inherited myopathies presenting with spinal rigidity.

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#### Abstract

**OBJECTIVES:** Inherited myopathies are major causes of muscle atrophy and are often characterized by rigid spine syndrome, a clinical feature designating patients with early spinal contractures. We aim to present a decision algorithm based on muscular whole body magnetic resonance imaging (mWB-MRI) as a unique tool to orientate the diagnosis of each inherited myopathy long before the genetically confirmed diagnosis.

**METHODS:** This multicentre retrospective study enrolled 79 patients from referral centres in France, Brazil and Chile. The patients underwent 1.5-T or 3-T mWB-MRI. The protocol comprised STIR and T1 sequences in axial and coronal planes, from head to toe. All images were analyzed manually by multiple raters. Fatty muscle replacement was evaluated on mWB-MRI using both the Mercuri scale and statistical comparison based on the percentage of affected muscle.

**RESULTS:** Between February 2005 and December 2015, 76 patients with genetically confirmed inherited myopathy were included. They were affected by Pompe disease or harbored mutations in RYR1, Collagen VI, LMNA, SEPN1, LAMA2 and MYH7 genes. Each myopathy had a specific pattern of affected muscles recognizable on mWB-MRI. This allowed us to create a novel decision algorithm for patients with rigid spine syndrome by segregating these signs. This algorithm was validated by five external evaluators on a cohort of seven patients with a diagnostic accuracy of 94.3% compared with the genetic diagnosis.

**CONCLUSION:** We provide a novel decision algorithm based on muscle fat replacement graded on mWB-MRI that allows diagnosis and differentiation of inherited myopathies presenting with spinal rigidity.

**KEY POINTS:** • Inherited myopathies are rare, diagnosis is challenging and genetic tests require specialized centres and often take years. • Inherited myopathies are often characterized by spinal

rigidity. • Whole body magnetic resonance imaging is a unique tool to orientate the diagnosis of each inherited myopathy presenting with spinal rigidity. • Each inherited myopathy in this study has a specific pattern of affected muscles that orientate diagnosis. • A novel MRI-based algorithm, usable by every radiologist, can help the early diagnosis of these myopathies.

**KEYWORDS:** Muscular diseases; Muscular dystrophies; Myopathies, structural, congenital; Spinal curvatures; Whole body imaging

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