Whole-body muscle magnetic resonance imaging in SEPN1-related myopathy shows a homogeneous and recognizable pattern

Cita: Hankiewicz, K., Carlier, R., Lazaro, L., Linzoain, J., Barnerias, C., Gomez, D., … Quijano-Roy, S. Whole-body muscle magnetic resonance imaging in SEPN1-related myopathy shows a homogeneous and recognizable pattern. *Muscle and Nerve, 52*(5), pp. 728-735. https://doi.org/10.1002/mus.24634

Asbtract

Introduction: The aim of this study was to delineate the spectrum of muscle involvement in patients with a myopathy due to mutations in *SEPN1* (SEPN1-RM). *Methods:* Whole-body magnetic resonance imaging (WBMRI) was used in 9 patients using T1-weighted turbo spin–echo (T1-TSE) sequences and short tau inversion recovery (STIR) in 5 patients. *Results:* Analysis of signal and volume abnormalities by T1-TSE sequences in 109 muscles showed a homogeneous pattern characterized by a recognizable combination of atrophy and signal abnormalities in selected muscles of the neck, trunk, pelvic girdle, and lower limbs. Severe wasting of sternocleidomastoid muscle and atrophy of semimembranosus were detected. Selective paraspinal, gluteus maximus, and thigh muscle involvement was also observed. The lower leg was less constantly affected. *Conclusions:* WBMRI scoring of altered signal and atrophy in muscle can be represented by heatmaps and is associated with a homogeneous, recognizable pattern in *SEPN1-RM,* distinct from other genetic muscle diseases.