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Format: Abstract

J Neurol. 2020 May 3. doi: 10.1007/s00415-020-09860-x. [Epub ahead of print]

Characteristic muscle signatures assessed by quantitative MRI in patients with Bethlem myopathy.

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Abstract

Using MRI, the main aim was to (1) map the pattern of muscle involvement by assessing fat fraction and (2) investigate frequency of target and sandwich signs in 42 muscles of patients with Bethlem myopathy (BM). Fifteen BM patients were included. Results were compared to findings in 8 healthy controls and 50 patients with four other types of muscular dystrophies. All muscles, except one, showed higher fat fraction in BM patients vs healthy controls ($p < 0.05$) with an overall proximal muscle affection, resembling a limb girdle-like pattern. In moderate patients, the specificity was 90% for the sandwich sign and 98% for the target sign. Sensitivity for both signs was 100%. Twelve BM patients had sandwich sign in other muscles than the vastus lateralis. Muscle strength correlated with fat fraction. Mean fat fraction in the psoas major was 39% in BM patients, which was considerably higher than in 3 of the 4 muscular dystrophy control diseases. The presence of signs in conjunction with severe affection of the psoas major muscle can serve as a diagnostic tool in BM. The high level of STIR lesions in muscles of BM patients warrants further investigations.

KEYWORDS: Bethlem myopathy; Quantitative MRI; Sandwich sign; Target sign; The psoas major muscle

PMID: 32363432 DOI: [10.1007/s00415-020-09860-x](https://doi.org/10.1007/s00415-020-09860-x)

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