Muscle Magnetic Resonance Imaging in Patients with Various Clinical Subtypes of LMNA-Related Muscular Dystrophy.

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Abstract in English, Chinese

BACKGROUND: LMNA-related muscular dystrophy can manifest in a wide variety of disorders, including Emery-Dreifuss muscular dystrophy (EDMD), limb-girdle muscular dystrophy (LGMD), and LMNA-associated congenital muscular dystrophy (L-CMD). Muscle magnetic resonance imaging (MRI) has become a useful tool in the diagnostic workup of patients with muscle dystrophies. This study aimed to investigate whether there is a consistent pattern of MRI changes in patients with LMNA mutations in various muscle subtypes.

METHODS: Twenty-two patients with LMNA-related muscular dystrophies were enrolled in this study. MRI of the thigh and/or calf muscles was performed in them. The muscle MRI features of the three subtypes were compared by the Mann-Whitney U-test. The relationship between the clinical and MRI findings was also investigated by Spearman's rank analyses.

RESULTS: The present study included five EDMD, nine LGMD, and eight L-CMD patients. The thigh muscle MRI revealed that the fatty infiltration of the adductor magnus, semimembranosus, long and short heads of the biceps femoris, and vasti muscles, with relative sparing of the rectus femoris, was the predominant change observed in the EDMD, LGMD, and advanced-stage L-CMD phenotypes, although the involvement of the vasti muscles was not prominent in the early stage of L-CMD. At the level of the calf, six patients (one EDMD, four LGMD, and one L-CMD) also showed a similar pattern, in which the soleus and the medial and lateral gastrocnemius muscles were most frequently observed to have fatty infiltration. The fatty infiltration severity demonstrated higher scores associated with disease progression, with a corresponding rate of $1.483 + 0.075 \times$ disease duration ($X$) ($r = 0.444, P = 0.026$). It was noteworthy that in six L-CMD patients with massive inflammatory cell infiltration in muscle pathology, no remarkable edema-like signals were observed in muscle MRI.
CONCLUSIONS: EDMD, LGMD and advanced-staged L-CMD subtypes showed similar pattern of muscle MRI changes, while early-staged L-CMD showed somewhat different changes. Muscle MRI of L-CMD with a muscular dystrophy pattern in MRI provided important clues for differentiating it from childhood inflammatory myopathy. The fatty infiltration score could be used as a reliable biomarker for outcome measure of disease progression.

KEYWORDS: Congenital Muscular Dystrophy; Emery-Dreifuss Muscular Dystrophy; LMNA; Limb-Girdle Muscular Dystrophy; Muscle Magnetic Resonance Imaging

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Conflict of interest statement