Facioscapulohumeral muscular dystrophy and dermatomyositis: a clinical case report

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INTRODUCTION

Facioscapulohumeral muscular dystrophy (FSHD) is an inherited myopathy with a prevalence of 4-10 cases per 100,000. Dermatomyositis (DM), an idiopathic inflammatory myopathy, has a prevalence of 1.97 to 21.5 occurrences per 100,000. Recent works have described patients with both FSHD and inflammatory myopathies.

MATERIALS AND METHODS

We report clinical, histological and muscle imaging data of a 24 years old man affected by FSHD (positive family history) who developed signs and symptoms of DM. The patient had scapular girdle weakness since eighteen years old, associated with mild facial weakness. The degree of muscle involvement remained stable over time. In March 2023 he presented an erythematous skin rash around the eyes, neck and phalangeal joints associated with increased CK (1562 UI/L) and weakness, with difficulty in walking and climbing stairs. A muscle MRI showed remarkable hyperintense signal on T2-weighted short-tau inversion recovery sequences (T2w-STIR) in many muscles of the upper girdle and the lower limbs. A muscle biopsy showed relevant perifascicular atrophy with hyperexpression of MHC-I on sarcolemma, pathological findings compatible with the diagnosis of DM. Screening for malignancies was negative. The patient underwent high-dose steroids and IVIG, with improvement of dermatological and muscle symptoms and lowering of the CK value to around 650 UI/L. A two-year follow-up muscle MRIs demonstrated marked reduction of the T2w-STIR positive muscle lesions.

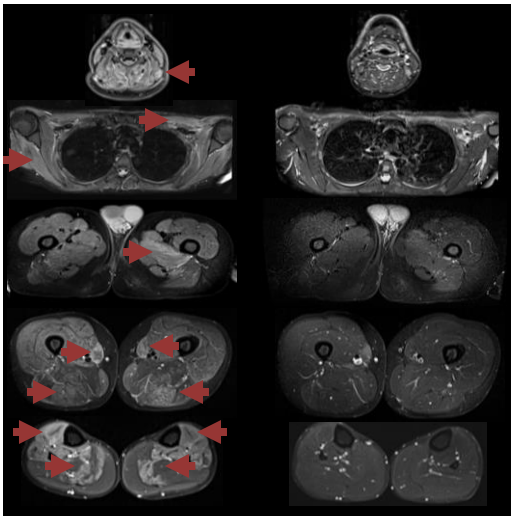

Figure 1 **Figure 2**

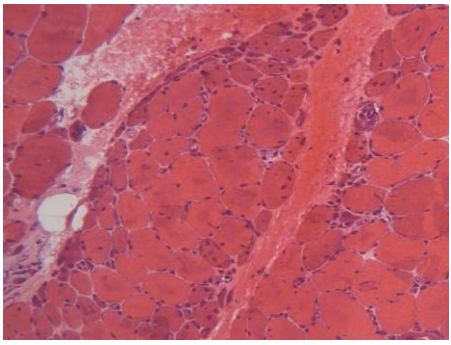

Figure 3

Figure 1. First muscle MRI (STIR sequence, 2023) shows diffuse hyperintensity in both upper and lower limb muscles (arrows).

Figure 2. Muscle MRI (STIR sequence, 2025) shows a marked reduction of T2w-STIR hyperintense lesions, indicating a positive treatment response following corticosteroid and IVIg therapy.

Figure 3. Muscle biopsy (2023), hematoxylin-eosin 10x, right vastus lateralis showing predominant perifascicular atrophy.

CONCLUSION

This case supports the hypothesis that the association between the two rare diseases may not be coincidental. Nonetheless, the differential response of muscle T2-weighted STIR hyperintense lesions to treatment with intravenous immunoglobulins and corticosteroids suggests the presence of distinct underlying pathogenic mechanisms.

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