



## SCLEROSIS: CLINICAL EXPERIENCE WITH ANTICD20 THERAPY

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### INTRODUCTION

The coexistence of Multiple Sclerosis (MS) and Facioscapulohumeral Muscular Dystrophy Type 1 (FSHD1) is extremely rare, with few cases described in the literature. MS is a chronic inflammatory demyelinating disease of the central nervous system (CNS), while FSHD1 is an autosomal dominant inherited muscular disorder caused by deletions of D4Z4 repeat units on chromosome 4q35. The simultaneous presence of these two conditions may lead to overlapping symptoms, complicating the clinical picture and potentially delaying diagnosis [1].

### CASE PRESENTATION

We report the case of a 43-year-old man with a previous diagnosis of FSHD1 who began to present new neurological symptoms, including paroxysmal headaches, balance disturbances, and visual changes. Neurological examination revealed asymmetric muscle atrophy, generalized weakness, scapular winging, and a positive Beevor sign. An initial brain MRI performed in 2021 showed a nonspecific hyperintense lesion, but follow-up imaging in 2023 and 2024 revealed multiple demyelinating lesions in both the brain and spinal cord. Cerebrospinal fluid analysis showed a type 2 oligoclonal band pattern, and visual evoked potentials were altered, consistent with a previous episode of optic neuritis.

### TREATMENT

- Based on clinical, radiological, and cerebrospinal fluid findings, a diagnosis of MS was confirmed.
- Given the high disease activity, treatment with Ublituximab, a high-efficacy anti-CD20 monoclonal antibody, was initiated [2].
- The therapy was well tolerated, and no adverse events were reported.
- The patient showed clinical stabilization and no relapses during follow-up.

### CONCLUSIONS

This case highlights the importance of maintaining a high index of clinical suspicion when patients with muscular dystrophies develop atypical neurological symptoms. The coexistence of MS and FSHD1 may reflect a shared genetic predisposition, further investigations will be necessary. The overlap between FSHD1 and MS can mask early signs of CNS involvement, leading to diagnostic and therapeutic delays. Our case adds to the limited body of literature on this rare association, emphasizing the need for a multidisciplinary diagnostic approach and supporting the use of high-efficacy therapies, such as Ublituximab, in complex clinical scenarios.

### Clinical and Diagnostic Timeline



#### REFERENCES

- [1] Iodice, R., et al. - Facioscapulohumeral muscular dystrophy (FSHD) and multiple sclerosis: a case report - *Acta Myol* - 2020 - 39(1) - 29  
[2] Steinman, L., et al. - Ublituximab versus Teriflunomide in Relapsing Multiple Sclerosis - *N Engl J Med* - 2022 - 387(8)-704



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