A case of facioscapulohumeral muscular dystrophy and myasthenia gravis with positivity of anti-Ach receptor antibody: a fortuitous association?

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Introduction

Facioscapulohumeral dystrophy (FSHD) is the third most common muscular dystrophy after Duchenne dystrophy and Myotonic dystrophy, inherited mostly as an autosomal dominant disease. It presented clinically with slowly progressive weakness and wasting of facial and limb muscles; bulbar, extraocular and respiratory muscles tend to be spared. Concomitant occurrence of FSHD and Myasthenia Gravis (MG) is not frequent; both are rare diseases with a prevalence of about 8 and 4 per 100,000 respectively.

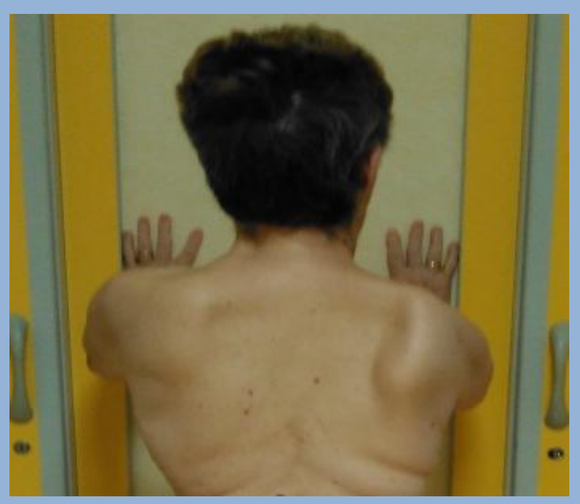
Case report

A 69-year-old woman had been diagnosed with FSHD confirmed by deletion analysis thirteen years ago and with arthritis in steroid treatment. She presented to our department with complaint of 20-day history of nasal timbre followed by dysphagia, diffuse weakness of all limbs with need of bilateral support during ambulation and in the last ten days appearance of binocular diplopia on vertical plan.

Neurological examination showed winging of scapula on right side, anserine gait possible with bilateral support, facial and limb-girdle muscle weakness and wasting with unilateral ptosis on the left, diplopia, dysphagia, dysarthria, depressed reflex. Serum creatine kinase values were high (414 UI/l).

Concentric needle electromyography showed findings suggestive of myopathy. **Repetitive nerve stimulation** produced a decremental response on orbicularis oculi muscle and trapezius suggestive of MG. Single fibre electromyography (**SFEMG**) was positive; **edrophomium test** was performed and dysarthria, ptosis and diplopia showed dramatic improvement. Serum anti-acetylcholine receptor (**AchR**) **antibody** level was marked elevated (16,01 nmol/l), confirming the diagnosis of MG. **Computerized tomography** of the thorax was normal. During hospital stay, the patient was exposed to intravenous immunoglobulin therapy and started on pyridostigmine with remarkable improvement.

Basale



Dopo test alla Neostigmina (0,5 mg ev)

Discussion & Conclusions

There are limited reports of concomitant occurrence of FSHD and MG in literature, but this association must be considered, to avoid a dangerous diagnostic delay, when any unusual changes in course of disease or development of unusual symptoms appear, as this case shows.

Our patient also suffered from arthritis and had elevated AchR antibody level, as the other cases described in literature, suggesting the possibility that immune mechanisms may be operative in FSHD. Both are infact rare diseases so a fortuitous association is unlikely. Moreover, AChR-abs have also been reported in patients with other genetic myopathies; theoretically, muscle fiber degeneration could cause autoinflammation that may lead to the breaking of immune tolerance to AchR and the generation of autoantibodies. This hypothesis is intriguing and widens the potential role of immunomodulation in these disease.

References:

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