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The Management of Dysferlinopathy in Physical Medicine and Rehabilitation - Case Report

Topic: Miscellaneous

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BACKGROUND

Dysferlinopathy is a group of autosomal recessive muscle diseases involving the gene coding for Dysferlin, a protein playing a Fundamental role in the mechanism of the muscle fiber membrane repair. The objective of this work is to review the role of physical medicine and rehabilitation in the management of this disease.

CASE DESCRIPTION

A 37-year-old female patient with a history of first-degree consanguinity and three similar cases in the family. She was referred to our department for the management of a myogenic syndrome, amyotrophy of the two lower limbs (MI) associated with disabling diffuse pain. Clinical examination revealed heaviness of the two IMs with amyotrophy making walking difficult without assistance, muscular weakness of the upper limbs was associated; the remainder of the examination objectified a myogenic syndrome predominating in the pelvic girdle with amyotrophy of both legs and bilateral steppageeles, ROTs were abolished. The electroneuromyogram confirmed myogenic damage, with normal sensitive and motor nerve conduction. Muscle biopsy and immunohistochemistry revealed profound dysferlin deficiency. Functional respiratory examination showed a restrictive syndrome.

DISCUSSION

Dysferlinopathy does not have an approved treatment, it is only symptomatic. Management must be tailored to the individual, including physical therapy. The patient benefited from a motor and functional rehabilitation protocol to optimize muscular performance, recover proprioception and prevent the onset of joint deformities. The protocol is based on physiotherapy, electrostimulation, occupational therapy, equipment, joint amplitude maintenance and stretching exercises to promote mobility and prevent contractures. Respiratory re-education sessions have been scheduled, with the aim of freeing the airways, improving thoracic ampliation and increasing lung volumes. Our patient showed a good clinical course, with reduced pain, partial recovery of walking and improved segmental and global muscle strength. Physical medicine treatment can be provided at any time, with the aim of educating the patient and maintaining his or her autonomy, as well as preventing complications and limiting deficiencies for good adaptation.

CONCLUSION

Two distinct phenotypes are associated with dysferlinopathies: distal Miyoshi myopathy and a form of limb-girdle muscular dystrophy LGMD2B. A multidisciplinary approach must be adopted from the moment of diagnosis, with rehabilitation as the first step, systematically combined with other therapeutic means, when considering a myogenic syndrome.

Statement of Disclosures

None

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