UNMASKING RESPIRATORY MUSCLE WEAKNESS DURING EFFORT IN MYOTONIC DYSTROPHY TYPE 1

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Background

Myotonic dystrophy type 1 (MD1), also known as Steinert's disease, is the most common monogenic autosomal dominant disorder affecting both skeletal and smooth muscles. Since treatment is symptomatic, early recognition of complications and timely, goal-oriented rehabilitation are essential.

Case report

A 50-year-old male with a confirmed diagnosis of MD1, under the care of a multidisciplinary team, was referred to a physical and rehabilitation medicine specialist for assessment of functional status and rehabilitation needs. At initial evaluation, hand muscle stiffness and weakness were observed. Following occupational therapy, the patient was referred for assessment of cardiorespiratory capacity based on a recommendation for respiratory rehabilitation. Isolated hypercapnia was identified despite otherwise normal pulmonary function tests, with the cause determined to be extrapulmonary—specifically, weakness of the respiratory muscles. The patient reported no signs of cardiorespiratory deconditioning. A 6-minute walk test showed normal performance (655 meters, minimum SpO_2 94%, expected blood pressure response to 140/90 mmHg). Maximal inspiratory pressure at rest (Pimax = 65 mbar) was within normal limits. Cardiopulmonary exercise testing, conducted using a RAMP protocol, revealed a VO_2max of 34.1 mL/kg/min (92% predicted), an oxygen pulse at 101% predicted, and a breathing reserve of 50.2%, all indicating preserved aerobic and ventilatory capacity. However, the VE/VCO_2 slope was elevated at 35.6, and VE/VCO_2 at the first ventilatory threshold was 27.9—both above normal, indicating reduced ventilatory efficiency. This finding, together with desaturation to 88% at peak exertion, confirmed exertional respiratory dysfunction since cardiopulmonary causes were excluded earlier. The patient was referred for targeted respiratory rehabilitation focusing on aerobic conditioning and strengthening of respiratory muscle function.

Conclusion

The aim of this case report is to highlight the importance of assessing cardiorespiratory capacity both at rest and during exertion to guide effective and personalized rehabilitation planning.

Keywords: myotonic, dystrophy, respiratory, muscles, weakness