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Poster Abstract: Clinical

Role of Respiratory Rehabilitation in Pompe Disease: A Case Report

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BACKGROUND

Pompe disease or glycogen storage disease type II is the accumulation of glycogen in muscle tissue due to a deficiency of lysosomal acid maltase. Today it is considered to be a rare disease, untreated until 2006. Prevalence is 1 case per 60,000 habitants, incidence is 1 case per 100,000 population per year. The disease causes severe cardiorespiratory and muscular disabilities. We report a case diagnosed in the Universitary Hospital Puerta del Mar (Cadiz, Spain), treated in a multidisciplinary way, with emphasis on the effect of pulmonary rehabilitation on increasing respiratory muscle strength.

CASE REPORT

A 22-year-old male with a history of hyperlipidemia and hypertension was admitted to the ICU for progressive generalized fatigability with acute respiratory failure requiring intubation and mechanical ventilation.

Clinical examination revealed a flaccid tetraparesis with proximal amyotrophy girdle, scapular winging, and weakness of the cervical flexor muscles. The overall muscular balance was 3/5. Weak tendon reflexes and flexor plantar response. Blood work had high LDH and CK levels. Spirometry: restrictive pattern with FVC 3.70 l, 2.90 l FEV1 FEV1/FVC 71.76. EMG study: pattern of repetitive discharges. Muscle biopsy suggestive of glycogenosis type II findings. Twenty-four-hour urine: elevated levels of glucose tetrasaccharide. Genetic study: carrier heterozygosity mutations c.2173 C> T (p.R725W) and c.2316 G> A (p.W772X).

The treatment was based on four aspects: (a) enzyme replacement therapy (alglucosidase alfa); (b) specific diet; (c) physical therapy; and (d) early respiratory rehabilitation, 1 session a day during 14 weeks. In order to: (a) facilitate bronchial drainage; (b) assist effective coughing; (c) encourage diaphragmatic and intercostal muscles; and (d) incentive breathing. After 6 months of treatment, complete motor recovery, weight gain, and muscle mass were achieved. The tracheal cannula was removed, spirometry parameters were normalized (FVC 4.98 l, l FEV1 3.8 FEV1/FVC 85.39.). For a few months he used BiPAP at night, now retired.

CONCLUSIONS

Respiratory failure is a serious threat to these patients. Specific treatment with alglucosidase alfa has been shown to modify the natural history, preventing muscle weakness and death from respiratory failure.[1] Pulmonary rehabilitation aims to prevent and resolve respiratory complications of diaphragmatic and intercostal muscle involvement and is associated with mechanical ventilation.[1] The improvement of spirometric parameters suggests that respiratory rehabilitation increases respiratory muscle strength, even in severe weakness.[2] Aerobic exercise helps improve motor function.[3] There are few reports in the literature of cases in which the enzyme therapy and respiratory therapy improve respiratory parameters to the point of being able to leave the intermittent or continuous mechanical ventilation (BiPAP®).

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