CASE REPORTS

Domiciliary Respiratory Muscle Training in Myotonic Dystrophy

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A 42-year-old man diagnosed with myotonic dystrophy experienced loss of respiratory muscle strength over a period of 6 months. We report the application of a domiciliary training program targeting both inspiratory and expiratory muscles. Maximal inspiratory and expiratory pressures, forced vital capacity, and forced mid-expiratory flow rate were measured 6 months before start of training, just before commencement of the program, and immediately after 12 weeks of training. Adherence to the program was satisfactory. Inspiratory muscle training was efficacious in increasing respiratory muscle strength. Expiratory muscle training, which made use of the Threshold PEP bronchial hygiene device incorporating an adapted flutter valve, was not efficacious in increasing maximal expiratory pressure or halting its loss. However, decreased obstruction of medium-caliber airways was observed with use of the device.

Key words: Myotonic dystrophy. Respiratory muscle training. Maximal respiratory pressures.

Introduction

Myotonic dystrophy, or Steinert disease, is an autosomal dominant disorder that develops slowly, with neuromuscular and systemic involvement. The main symptoms are muscular weakness, myotonia, gonadal atrophy, cataracts, cardiac arrhythmias, and restrictive pulmonary impairment. Pulmonary involvement is usually secondary to muscle weakness and develops along with diminished thoracic mobility and respiratory muscle strength.

Respiratory muscle training has been shown to be effective for improving muscle function in some diseases that cause muscle weakness. Training also facilitates ventilator weaning in postoperative patients. Moreover, muscle weakness in general and respiratory muscle weakness in particular often lead to respiratory complications in a variety of circumstances. However, the effectiveness of muscle training has never been investigated in the context of myotonic dystrophy. We report the case of a patient with this disease for whom tailored respiratory muscle training led to clear improvement in symptoms and lung function.

Case Description

A 42-year-old man (body mass index 23 kg/m²) diagnosed with myotonic dystrophy when he was 30 years old presented signs of muscle weakness in hands and had slight difficulties walking. Shortness of breath appeared with moderate exercise but there was no evidence of orthopnea or hypoventilation. We reviewed lung function tests performed 6 months before the decision to begin respiratory muscle training and at the...
start and end of training. The lung function tests revealed slight restrictive abnormalities and maximum respiratory pressures within reference values. At the time of the visit to start therapy (6 months after the first visit), the lung function study also revealed a slight decline in respiratory muscle strength but no change in basic spirometric parameters (Table).

The patient was advised to undertake a respiratory muscle training program, taught in the hospital and continued at home. That program included the following components: inspiratory muscle training (IMT) with a specific device (Threshold IMT, Respironics, Cedar Grove, New Jersey, USA) which provided an inspiratory resistance between 7 and 20 cm H₂O and expiratory muscle training with a device able to provide positive expiratory pressure (PEP) in the form of a resistive load ranging from 4 to 20 cm H₂O (Threshold PEP, Respironics). The home program included inspiratory and expiratory exercises lasting 5 minutes each twice daily (morning and afternoon) 3 times a week for 12 weeks. The resistive load on the Threshold IMT device was increased gradually, usually between 5% and 7% every 2 weeks, rising from 15% to 60% of maximum inspiratory pressure. The resistive load for expiratory muscle training increased 5% every 2 weeks until the 10th week, after which there was no change, for a total increase of 10% to 30%. The patient received verbal and written instructions to contact the neurologist or the physiotherapist in case of a problem. The physiotherapist called the patient weekly for 12 weeks to check that the devices were working and to ask about the patient’s condition. At the end of the training period the physical examination and lung function tests were repeated.

The new study showed that inspiratory muscle strength had increased but that expiratory muscle strength and spirometric values remained unchanged. Blood gases were also unchanged and the patient complained of slight shortness of breath with exercise (Table).

**Discussion**

We report the case of a man with myotonic muscular dystrophy who experienced gradual loss of muscle strength and mild respiratory symptoms. Clinical and functional improvement was observed, though midexpiratory flow was the only spirometric variable that increased. Improvement was the result of a tailored training program that has not been described to date in the literature on this type of patient.

**REFERENCES**


### Lung Function During Clinical Follow-up

<table>
<thead>
<tr>
<th></th>
<th>6 Months Before Training</th>
<th>At Start of Training</th>
<th>After Training</th>
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<tbody>
<tr>
<td>FVC, L (%)</td>
<td>4.9 (84)</td>
<td>5.1 (88)</td>
<td>4.9 (84)</td>
</tr>
<tr>
<td>FEV₁, L (%)</td>
<td>3.7 (84)</td>
<td>3.7 (63)</td>
<td>3.7 (83)</td>
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<tr>
<td>FEV₁/FVC, %</td>
<td>76</td>
<td>76</td>
<td>76</td>
</tr>
<tr>
<td>FEF₂₅₋₇₅ , L-s⁻¹ (1)</td>
<td>3.17 (74)</td>
<td>2.49 (59)</td>
<td>3.10 (73)</td>
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<tr>
<td>P pł (cm H₂O (1)</td>
<td>100 (100)</td>
<td>61 (62)</td>
<td>90 (91)</td>
</tr>
<tr>
<td>P pł,max (cm H₂O (1)</td>
<td>97 (68)</td>
<td>67 (47)</td>
<td>51 (36)</td>
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<td>pH</td>
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<tr>
<td>PaO₂, mm Hg</td>
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<tr>
<td>PaCO₂, mm Hg</td>
<td>45</td>
<td>39</td>
<td>45</td>
</tr>
</tbody>
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*PEF₂₅₋₇₅, indicates forced midexpiratory flow; FEV₁, forced expiratory volume in 1 second; FVC, forced vital capacity; P pł,max, maximal inspiratory pressure; P pł,max, maximal expiratory pressure.*


