

Domiciliary Respiratory Muscle Training in Myotonic Dystrophy

Guilherme Augusto de Freitas Fregonezi, Vanessa Regiane Resqueti, and Pere Casan

Departamento de Neumología, Hospital de la Santa Creu i Sant Pau, Facultad de Medicina, Universitat Autònoma de Barcelona, Barcelona, Spain.

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 midexpiratory 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.¹ 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

Entrenamiento muscular respiratorio domiciliario en la distrofia muscular miotónica

Se describe el caso de un varón de 42 años de edad con diagnóstico de distrofia muscular miotónica e historia de disminución de la fuerza muscular respiratoria en los últimos 6 meses. Relatamos la aplicación de un programa de entrenamiento muscular respiratorio (inspiratorio e espiratorio) domiciliario. En los 6 meses previos al entrenamiento, justo antes del comienzo del programa e inmediatamente después de 12 semanas de su inicio se estudiaron la presión inspiratoria máxima, la presión espiratoria máxima, la capacidad vital forzada y el flujo mesoespiratorio forzado.

El cumplimiento del programa fue satisfactorio. El entrenamiento muscular inspiratorio se mostró efectivo para aumentar la fuerza muscular respiratoria. El entrenamiento muscular espiratorio utilizando la válvula adaptada de higiene bronquial Threshold PEP®, adaptada para el entrenamiento, no fue eficiente para aumentar la presión espiratoria máxima y/o frenar su disminución. Sin embargo, con la utilización de la válvula Threshold PEP® se observó la disminución de la obstrucción de las vías aéreas de medio calibre.

Palabras claves: *Distrofia muscular miotónica. Entrenamiento muscular respiratorio. Presiones respiratorias máximas*

diseases that cause muscle weakness.²⁻⁴ Training also facilitates ventilator weaning in postoperative patients.⁵

Moreover, muscle weakness in general and respiratory muscle weakness in particular often lead to respiratory complications in a variety of circumstances.^{6,7} 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

GA de Freitas Fregonezi is a doctoral fellow of CNPq-Brasil (process 2000-05/01-4).

Correspondence: Dr. G.A. de Freitas Fregonezi
Departamento de Neumología, Hospital de la Santa Creu i Sant Pau
Antoni Maria Claret, 167. 08025 Barcelona, España
E-mail: gfrezonezi@msn.com

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TABLE
Lung Function During Clinical Follow-up*

| | 6 Months Before Training | At Start of Training | After Training |
|--|--------------------------|----------------------|----------------|
| FVC, L (%) | 4.9 (84) | 5.1 (88) | 4.9 (84) |
| FEV ₁ , L (%) | 3.7 (84) | 3.7 (83) | 3.7 (83) |
| FEV ₁ /FVC, % | 76 | 73 | 76 |
| FEF _{25%-75%} , L·s ⁻¹ (%) | 3.17 (74) | 2.49 (59) | 3.10 (73) |
| PI _{max} , cm H ₂ O (%) | 100 (100) | 61 (62) | 90 (91) |
| PE _{max} , cm H ₂ O (%) | 97 (68) | 67 (47) | 51 (36) |
| pH | 7.39 | 7.41 | 7.41 |
| PaO ₂ , mm Hg | 84 | 86 | 82 |
| PaCO ₂ , mm Hg | 45 | 39 | 45 |

*FEF_{25%-75%}, Indicates forced midexpiratory flow; FEV₁, forced expiratory volume in 1 second; FVC, forced vital capacity; PI_{max}, maximal inspiratory pressure; PE_{max}, maximal expiratory pressure.

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 41 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.

Respiratory muscles can be affected by myotonic muscular dystrophy even without the observable impairment of muscles in the upper or lower extremities.⁶ This pattern is very similar to that described recently for Duchenne muscular dystrophy,⁸ although it not the most common one, as respiratory muscle decline generally follows loss of muscle strength in the extremities.⁹⁻¹¹ Our patient's clinical picture corresponded to the one described in the literature, although clinical and functional decline was observed in the last 6 months before training.

Respiratory muscle weakness implies loss of coughing force and effectiveness, one of the most immediate consequences being more frequent lung infections. In addition, the involvement of inspiratory function eventually causes hypercapnia, which may develop soon or be delayed. In both cases respiratory muscle involvement culminates in respiratory failure.^{12,13}

Although satisfactory results were observed in relation to inspiratory muscles and midexpiratory flow, it must be recognized that the most important limitation in this case was the commercial valve used for expiratory muscle training. Because the load could not be increased beyond 20 cm H₂O, it was impossible to halt the loss of expiratory muscle strength. Also important is the fact that training did not lead to improved forced vital capacity or forced expiratory volume in 1 second in spite of satisfactory improvement in midexpiratory flow. The reason may be related to the low training load provided by the commercial valve, which was designed to improve bronchial hygiene and induce a huff cough. There are no valves available on the market designed expressly for expiratory muscle training. Finally, another important finding was the improvement in PaCO₂ after 12 weeks of training. This observation suggests that improvement might have been related to a switch to a more efficient breathing pattern, although we did not measure the relevant variables to confirm that.¹⁴

Our patient's increase in inspiratory muscle strength (36%) allows us to predict slower functional decline as well as slower progression to respiratory failure.^{4,15} Perhaps if we had applied loads greater than 60% of maximum inspiratory pressure, the results would also have led to improved forced vital capacity. Improvement in midexpiratory flow is interpreted to indicate greater facility in mobilizing secretions and greater availability of peripheral airflow. Because the program is home based, its simplicity and applicability allow us to propose it for certain patients in order to objectively evaluate the effects of short- and long-term muscle training of this type.

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