Response

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Response: Precise Pulmonary Function Evaluation and Management of a Patient With Freeman-Sheldon Syndrome Associated With Recurrent Pneumonia and Chronic Respiratory Insufficiency (*Ann Rehabil Med* 2020;44:165-70)

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We appreciate the writer's interest and comment on the recently published article "Precise Pulmonary Function Evaluation and Management of a Patient With Freeman-Sheldon Syndrome Associated With Recurrent Pneumonia and Chronic Respiratory Insufficiency" [1]. The diagnosis of Freeman–Burian syndrome (FBS), a novel term replacing Freeman–Sheldon syndrome, is based on physical characteristics, especially craniofacial abnormalities, such as whistling face, microstomia, prominent nasolabial folds, and H- or V-shaped chin defect [2]. Previous reports had shown that mutations in the embryonic myosin heavy chain (MYH3) have strong correlation with FBS [3], while it may be non-diagnostic [2]. As the writer has pointed out, we did not focus on mentioning the diagnostic criteria of FBS; however, the patient in the study meets the diagnostic criteria. The patient had characteristic facial abnormalities of FBS as shown in figure and a gene defect in MYH3. For additional information on the diagnostic criteria for FBS, please refer to the paper authored by Poling et al. [2].

As per the writer's comment, it is essential to deliberate more regarding the cause of the respiratory failure. This leads to the query regarding the cause of chronic respiratory failure of restrictive pulmonary disease. In FBS patients, as in the case of most patients with neuromuscular disease, the muscles involved in respiration are weakened [4]. It is notable that problems with the nervous system can cause the respiratory muscles to fail to function [5]. Alternately, severe scoliosis can be the problem [6].

As mentioned above, there can be a plethora of potential causes of respiratory failure in FBS patients. Thus, it is challenging to pinpoint the main cause of respiratory failure in patients with FBS. This patient clearly has a

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weakness of the inspiratory and expiratory muscles, with a vital capacity of under 20% of the normal predictive value, a maximal inspiratory and expiratory pressure under 30% of the normal predictive values on repeated evaluations. We first considered the possibility that patients with FBS might have weakness of the respiratory muscles since there was a report that the weakening of intercostal muscles in patients with FBS is observed [7]. However, it is difficult to say this with certainty from a single case report without much needed details on proper pulmonary function tests or records of pulmonary rehabilitation at the onset of respiratory failure. Notably, no literature regarding accurate pulmonary functions of FBS to confirm the weakness of respiratory muscle strength can be found. The clinical course of our patient was similar to the previous report of FBS with respiratory failure with scoliosis [8]. Hence, we concluded that the main cause of the respiratory failure might be severe scoliosis.

However, as the letter pointed out, the normal muscle fibers were replaced by white fibrous tendinous-like tissue resulting in decreased muscle strength in FBS. This suggests that the respiratory muscles might be affected in some cases of severe FBS. We agree with the letter's opinion and want to explore the possibility of weakened respiratory muscles being the cause of respiratory failure in FBS. To prove this, it is necessary to obtain the pulmonary functions of a number of FBS longitudinally.

Additionally, while the letter emphasized the importance of physical exercise, it is impossible to impede the progression of pathologic muscle weakness including that of respiratory muscles by physical exercise alone in patients with most neuromuscular disorders [9]. Mechanical ventilation support and cough augmentation techniques are essential for those patients to maintain their cardiopulmonary function and to improve the quality of life [10].

In our case, the patient showed hypercapnia without using ventilator in a few hours indicating a need for ventilatory support. However, from a different viewpoint, the patient had several hours in a day to stay without a ventilator before showing hypercapnia. Additionally, after learning how to perform cough augmentation techniques, he had not suffered from pneumonia which had repeatedly occured. Mechanical ventilation and cough augmentation techniques had a significant impact on his health and quality of life.

In summary, it would be necessary to conduct a regular assessment of pulmonary function in FBS patients. If respiratory failure due to severe respiratory muscle weakness is observed, physicians should consider using mechanical ventilation and educating an cough augmentation techniques. Further studies are needed to determine if the patients with FBS have additional causes of respiratory failure other than severe scoliosis.

CONFLICT OF INTEREST

No potential conflict of interest relevant to this article was reported.

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