Letter to the Editor

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Letter: 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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Dear editor,

We read with great interest the case report by Park et al. [1], titled "Precise Pulmonary Function Evaluation and Management of a Patient with Freeman-Sheldon Syndrome Associated With Recurrent Pneumonia and Chronic Respiratory Insufficiency". It is encouraging to see this exquisitely rare condition, now referred to as Freeman–Burian syndrome (FBS), accurately identified in the literature and the critical issue of restrictive pulmonary disease addressed in this context [1,2]. Unfortunately, this article is unclear to some extent, apparently resulting from the authors' omission of relevant literature. In discussing the major clinical features of FBS, they list quite common features, including some from the diagnostic criteria [1,3]. However, they do not state the clinical diagnostic criteria [1,3]. They place a significant emphasis on distal extremity contractures, which are a non-diagnostic finding in FBS and common in several syndromic and non-syndromic entities [1–3]. Not explicitly stating the diagnostic criteria can confuse readers who are unfamiliar with FBS [2].

While FBS has had several classifications since its first description in 1938, the most reasonable seems to be as a complex congenital myopathic craniofacial syndrome [4]. Since the features of distal arthrogryposis are not required for diagnosis, FBS cannot be cogently classed as a distal arthrogryposis syndrome, though many authors and clinicians continue to do so [4].

While the authors do refer to the weakness of respiratory muscles, they, like several authors, place a greater importance on the orthopedic deformities [1]. In the syndrome, orthopedic deformities are a symptom and not a cause [2]. In FBS, white fibrous tendinous-like tissue replaces histologically normal muscle fibers to varying degrees [2]. This is correlated with *in vitro* molecular myophysiology observations showing problems with the metabolic process for contraction and extreme muscle stiffness that reduces the muscular work and power [2]. Previous authors have observed non-functioning inter-

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costal muscles [5] and in FBS, the potential presence of ineffective intercostal muscles—combined with severe abnormal spinal curvatures—can lead to a reduced intrathoracic volume, impaired thoracic cage compliance, and restrictive pulmonary disease [2].

Finally, regarding the treatment of the patient's restrictive pulmonary disease, the authors describe invasive and non-invasive positive pressure ventilation, without mentioning the importance of functional pulmonary rehabilitation in these patients [1]. As a myopathy, it is essential to encourage these patients to maintain an active lifestyle and exercise to prevent or limit secondary effects, such as the progression of spinal curvatures and other orthopedic deformities [2]. There is growing evidence supporting the use of exercise in treating several conditions, and the evidence-base using exercise as a treatment for pulmonary diseases is especially strong. While the patient did well [1], the use of positive-pressure ventilation, whether invasive or non-invasive, is not benign. Nonetheless, in addition to importantly contributing to the growing body of evidence for pulmonary pathology in FBS, this article also illustrates the perils of describing a rare condition.

CONFLICT OF INTEREST

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

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