

Plenary Abstract

Early Recognition of Pompe Disease by Respiratory Muscle Signs and Symptoms

P. Young* and M. Boentert

Department of Sleep Medicine and Neuromuscular Disorders, University of Münster, Münster, Germany

Ventilatory aspects in adults with Pompe disease have a major impact on clinical course and quality of life. Treatment with non-invasive ventilation is state of the art for these patients. However, it is well known that the onset of ventilatory symptoms does not strictly correlate with the degree of proximal skeletal muscle involvement. Early signs and symptoms indicating ventilatory insufficiency are often discrete and may be overlooked. Besides the clinical impact there is enough evidence that the ventilatory insufficiency, as in other neuromuscular disorders, affects quality of sleep also in early stages due to early hypercapnic hypoventilation during the rapid-eye-movement (REM) sleep. There is a significant correlation of fatigue, daytime sleepiness and bad sleep in adult patients with Pompe disease (Boentert et al. [1]). This knowledge may help to recognise adult patients with Pompe disease by the signs and

symptoms indicating ventilatory insufficiency such as daytime sleepiness, fatigue, dyspnoea, orthopnoea, and paradoxical breathing. Further hypercapnic phases during REM sleep when performing sleep recordings with polysomnography in combination with transcutaneous capnometry are strong hints for early stages of hypoventilation in these patients. For that reason, patients suffering from proximal muscle weakness and wasting in combination with hypercapnic hypoventilation should be tested for enzyme activity of acidic glucosidase in the early steps of differential diagnosis.

REFERENCE

- [1] Boentert, M., et al. (2015) Sleep-related symptoms and sleep-disordered breathing in adult Pompe disease. *Eur J Neurol*, 22: 369–e27. doi: 10.1111/ene.12582

*Correspondence to: P. Young, University of Münster, Department of Sleep Medicine and Neuromuscular Disorders, Albert-Schweitzer Campus 1, 48149 Münster, Germany. E-mail: young@uni-muenster.de.