

# Daytime non-invasive ventilation via intermittent abdominal pressure in a patient with Pompe disease

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## Introduction

Pompe disease (PD) is a rare glycogen storage disorder that leads to progressive myopathy of respiratory muscles causing nocturnal hypoventilation, diurnal hypercapnia and sleep disordered breathing (SDB) [1]. Non-invasive ventilation (NIV) is indicated to improve gas exchanges, sleep outcomes and quality of life (QoL) [2, 3].

## Case report

A 22-year-old male university student affected by late onset PD (LOPD) came to our observation on January 2019. He was treated with enzyme replacement therapy (ERT) by the age of 11 and he started NIV at the age of 15 after diagnosis of moderate obstructive sleep apnoea with associated nocturnal hypoventilation.

At the first examination he presented with dorso-lumbar scoliosis, severe thinness and diffuse muscular weakness, complaining daytime sleepiness and morning headaches.

He underwent sniff nasal inspiratory pressure (SNIP: 30 cmH<sub>2</sub>O), spirometry (FVC 1,10 L, 21% of predicted; FEV<sub>1</sub> 1 L, 22%), arterial blood gas analysis (ABG) and type3 polysomnography (PSG) during NIV. QoL was evaluated via McGill QoL Questionnaire. NIV was indicated to be used during afternoon rest too.



Fig. 1. Patient wearing IAPV

After 1 month patient complained persistent daytime symptoms. He reported not daytime NIV use due to study related issues. PSG: no residual events.

**Intermittent abdominal pressure ventilator (IAPV)** was then initiated (Fig 1). After IAPV (PBelt 50) one hour trial, ABG (Table 1) and tidal volumes improved substantially. Diaphragmatic ultrasound were performed (Table 2).

At 3 months follow up, patient reported improved symptoms. PSG: no residual events. One hour IAPV use confirmed improvement in ABG and volumes. Diaphragmatic ultrasound and QoL questionnaire were also repeated.

Table 1. Respiratory gas exchanges

	Baseline (T0)	After 1 month (T1)		After 3 months (T2)	
	SB	SB	IAPV (1 h)	SB	IAPV (1 h)
pH	7.41	7.40	7.47	7.43	7.45
pCO <sub>2</sub>	51	51	39	50	44
pO <sub>2</sub>	87	85	94	89	97
HCO <sub>3</sub> <sup>-</sup>	32,3	31.6	28.4	33.2	30.6
BE	6.7	6.8	4.7	8.9	6.6

SB, spontaneous breathing; BE, base excess

Table 2. Ultrasound evaluation of diaphragmatic thickness

	T1	T2
Left side thickness (cm)	0,15	0,11
Right side thickness (cm)	0,13	0,21
Right side excursion during respiration at tidal volume (cm)	1,06	0,9
Right side excursion during a maximal respiratory act (cm)	1,79	1,79
Right side excursion during a sniff maneuver (cm)	1,22	1,38

## Conclusions

In LOPD, IAPV can be a successful daytime NIV tool, allowing good control of daytime symptoms, diaphragm thickness and QoL.

## References

1. Boentert M, Dräger B, et al. Sleep-Disordered breathing and effects of noninvasive ventilation