Late respiratory failure in Jeune syndrome

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Received: 30 July 2009 / Accepted: 7 August 2009 / Published online: 27 August 2009
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Sir,

With great interest, we read the excellent paper of De Vries et al. on their experience with Jeune syndrome (JS) [1]. Based on their observations, the authors conclude that respiratory condition in JS improves with age: in the 10 children surviving the first years of life, only two needed supplementary oxygen therapy beyond the age of two and none suffered from severely impaired exercise intolerance. However, we doubt that these findings allow the conclusion of absence of major respiratory problems later in life.

Recently, a 9-year-old girl with JS was referred to us. She had suffered from severe respiratory problems in her first years of life and had needed supplementary oxygen until the age of 3.5. Since then, her respiratory condition

had improved and at the age of 8 years, her exercise tolerance was considered as only mildly impaired. However, present history taking revealed severe fatigue, important sleeping disturbances, fear to go to sleep, nightmares, awakening difficulties and headache in the morning, diminished exercise tolerance, deteriorating school results, depression, and emotional instability. Spirometry showed a forced vital capacity of 28% of predicted. Sleep polygraphy revealed frequent arousals, a decreased oxygen saturation (90–93%), and a hypercapnia (pCO₂ up to 6.8 kPa). Despite her restrictive respiratory condition, noninvasive ventilation by facemask resulted in normocapnia during sleep. Subsequently, most of the patient's complaints dissolved.

This case illustrates that apparent initial recovery of respiratory function in Jeune syndrome does not exclude respiratory failure later in life. De Vries et al suggest annual respiratory follow-up by spirometry. However, the exact meaning of spirometry in the follow-up of Jeune syndrome still has to be determined. We believe that, just like in patients with neuromuscular diseases, patients with severe restrictive respiratory diseases should be evaluated regularly for signs and symptoms of sleep-related hypoventilation [2, 3].

Conflict of interest Regarding the subject of this letter, none of the authors has any conflict of interest or any relevant financial relationship to report.

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