

The impact of obesity on sleep, pulmonary and chest wall restriction in Osteogenesis Imperfecta: a pilot study.

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Introduction: Osteogenesis Imperfecta (OI) is characterised by brittle bones, severe skeletal deformities, low sleep quality, and restricted breathing. We aimed to distinguish how disease and obesity affect these results.

Methods: according to BMI, we considered four groups of peer adults (median age: 35.0 years): 13 subjects affected by moderate or severe OI without obesity (OI_{no}), 14 affected by moderate or severe OI with obesity (OI_{ob}), 10 without obesity not affected by OI (OB) and 10 without obesity not affected by OI.

Results: obstructive sleep apnoea syndrome was diagnosed in 4 OI_{no} (30%) and 9 OI_{ob} (64%). Restricted lung pattern (z-score of total lung capacity < - 1.64) was diagnosed in 10 OI_{no} (77%); 9 OI_{ob} (65%), and 1 OB (10%) subjects. In the seated position, OI_{ob} breathed with reduced tidal volume and higher respiratory rate, resulting in hypoventilation. Both OI_{no} and OI_{ob} were characterised by rapid and shallow breathing and lower ribcage expansion (negative in 3 (23%) OI_{no} and 3 (21%) OI_{ob}). In the supine position, the ventilatory pattern was similar among the four groups, while both OI_{no} and OI_{ob} were characterised by reduced ribcage contribution, which was negative in 6 (46%) OI_{no}, 11 (78%) OI_{ob} and 1 (10%) OB.

Conclusions: this is a pilot study on a small sample, the findings and conclusions apply only to this study population. The preliminary results suggest that in subjects with moderate or severe OI per se implies (1) a 30% prevalence of obstructive sleep apnoea syndrome, (2) a restricted lung pattern, (3) a lower ribcage expansion, and (4) rapid and shallow breathing in the seated position. The additional impacts of obesity on OI seem to determine (1) a higher incidence of obstructive sleep apnoea syndrome, (2) hypoventilation in the seated position, and (3) a higher incidence of paradoxical breathing lying supine. Reversing obesity in OI is even more challenging as knowledge of the diet and the physical activity suited for these patients is still scarce.

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