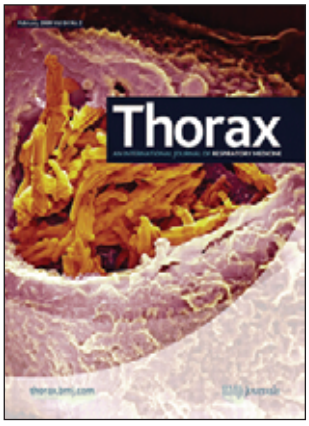
Obstructive sleep apnoea and aortic dilatation in Marfan's syndrome

Craniofacial abnormalities and greater pharyngeal collapsibility due to abnormal connective tissue suggest the possibility of an increased prevalence of obstructive sleep apnoea (OSA) in Marfan's syndrome, but the actual prevalence is uncertain.



Materials and methods

A total of 61 patients with Marfan's syndrome and 26 matched control subjects were recruited through an advertisement in the Oxford Radcliffe Hospitals, Oxford, UK. A two-channel polygraphic system with nasal pressure registration and oximetry was used. Aortic root diameter was measured by echocardiography.

Results

Of the 61 patients, 32.8% (apnoea/hypopnoea index (AHI) >5) or 18% (AHI>15) had some sleep disordered breathing (SDB), while controls showed OSA in 11.5% and 0%, respectively. AHI was significantly associated with aortic root diameter ($r=0.50$, $p<0.01$). Mean aortic root diameter was significantly greater in patients with (4.5±0.6 cm) than without OSA (3.7±0.6 cm; $p<0.01$).

Conclusion

The prevalence of OSA is considerably higher in patients with Marfan's than in matched controls. Moreover, OSA may be a risk factor for aortic root dilatation in Marfan's syndrome.

Message

Sleep disordered breathing is common in Marfan's syndrome, and could be related to outcome in these patients. Sleep studies are warranted in the clinical assessment.

Competing interests
None declared.

Original article
Kohler M, Blair E, Risby P, et al. The prevalence of obstructive sleep apnoea and its association with aortic dilatation in Marfan's syndrome. Thorax 2009; 64: 162–166.

Editorial comment

This observational and descriptive study in 61 patients with Ghent criteria-positive Marfan's syndrome illustrates the very high prevalence of SDB in Marfan's syndrome. In an earlier study, however, CISTULLI and SULLIVAN [1] reported a higher prevalence, which may be explained by a selection bias due to a relatively small number of randomly recruited patients. The present study also included a higher percentage of females, known to have a lower prevalence than men. In a questionnaire study, we found that 27% of Marfan's patients had features and symptoms of sleep apnoea, close to the data in the present study. A limitation of the current study is that the authors pretend to have performed an objective evaluation by a home sleep study, while having used a level 4 polygraphy. This could have led to underestimation of the real AHI and of the reported prevalence of SDB.

Marfan's patients have been considered before to have a high risk of sleep apnoea, due to high arched palate, nasal obstruction and retrognathia. The authors of the present study, however, found that Marfan's patients were not different from control subjects regarding the frequency of a retrognathic facial profile, mean neck circumference, cricomental distance or Mallampati score. Therefore, it seems that the higher prevalence of OSA found in patients with Marfan's syndrome is more likely to be the result of increased upper airway collapsibility rather than of craniofacial abnormalities. This also suggests that the relationship between SDB and Marfan's is still poorly understood.

The incidence of Marfan's syndrome is estimated to be 2-3 per 10,000, which makes it a quite common disease. However, the number of patients effectively treated for SDB is small, and the importance of SDB in Marfan's syndrome seems neglected. Therefore, a detailed medical history, including questions on snoring, apnoeas and sleepiness, together with systematic screening for SDB in Marfan's is warranted. Simply assessing a patient's subjective sleepiness is not useful, since, unexpectedly, the degree of subjective sleepiness was not related to the severity of SDB. The impact of treating SDB in Marfan's is substantial. In this study, it was found that the mean aortic root diameter is strongly related to the AHI, while a study in the general population has reported a high prevalence of SDB in patients with aortic dissection. In two case reports it has been demonstrated that continuous positive airway pressure (CPAP) ventilation and bilevel CPAP can attenuate aortic root dilatation [2, 3]. These data are hopeful, but a randomised controlled trial on the effects of CPAP on aortic root diameter in patients with SDB and Marfan's is required to prove this relationship. Also, new studies based on polysomnography in well described Marfan's patients are welcome, given the high prevalence of SDB in this population and the life-threatening consequences of this clinical problem.

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References

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