



Understanding the links between snoring, OSA and aortic root pathologies in Marfan syndrome

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Marfan syndrome (MFS) is an autosomal dominant multi-systemic genetic disorder that affects the connective tissues of the body. For individuals with this disorder, aortic root dilatation and subsequent dissection are the most common life-threatening manifestations. Obstructive sleep apnoea (OSA) is also highly prevalent in individuals with MFS and its presence has been shown to be a risk factor for aortic root dilation and replacement [1, 2]. More recently, self-reported snoring, which is commonly observed in individuals with OSA, has been shown to also be associated with enlargement of the aorta in patients with MFS.

In the current issue, Sowho and colleagues [3] expand on this body of work and examine the relationship between quantitative measures of snoring, the presence/absence of OSA and aortic root diameter in a cohort of patients with MFS. During an overnight in-laboratory polysomnogram, snoring was captured using a high fidelity sound pressure level meter and was defined as breaths in which the peak sound during inspiration was ≥ 40 dB. The authors then calculated the proportion of snoring during sleep that met or exceeded this level (which was termed the snoring breath %). The key finding of this novel investigation was that snoring is associated with both the presence of OSA as well as increasing aortic diameter in MFS patients. We believe that the current findings are important for the field as they confirm the importance of screening for snoring in individuals

with MFS. As the authors note, the development of home-based technologies (i.e. smart phones) could help facilitate identification of those at risk of OSA.

The current paper also critically signposts opportunities for future research exploring the impact of snoring and OSA on the MFS community. First, it is unclear whether it is snoring per se or OSA that drives the observed increases in aortic diameter. Whilst not conclusive, the association between the snoring breath % and aortic root diameter exclusively in OSA patients in the current study suggests an important potential role for OSA pathophysiology contributing above and beyond the noise of snoring. It causes us to question whether the relationship between snoring and aortic diameter also exists (and if there is a dose dependency) when using other snoring intensity thresholds [4, 5] [i.e. > 50 dB, 70 dB or overall intensity]? This work highlights the importance of future studies aimed at teasing apart the effects of snoring versus the effects of OSA on aortic diameter. Second, given that other novel PSG-derived metrics such as the hypoxic burden [6] have been shown to be predictive of cardiovascular disease, we wonder whether such metrics (or other novel PSG-derived metrics; i.e. flow-drive [7]) may be stronger predictors of the changes in aortic diameter. Lastly, research will also be required to test whether resolution of snoring (through interventions such as surgery or continuous positive airways pressure, CPAP), both in those with and without OSA, is capable of reversing the impacts on aorta dimensions. Certainly, there is some evidence albeit limited, that treatment of OSA with CPAP has been associated with attenuation of aortic root dilatation [8, 9].

The authors are to be congratulated on their work which highlights a clear need for an awareness of MFS patients' snoring status. The next phase of work will need to delineate the specific individual contributions of both snoring and OSA to worsening aortic diameter. Understanding these contributions will act as a platform for meaningful intervention studies aimed at slowing or reversing aortic pathologies in MFS patients.

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