Reviewer’s report

Title: Fatigue and disturbances of sleep in patients with Osteogenesis imperfecta - a cross-sectional questionnaire study

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Reviewer: John Gjevre

Reviewer's report:

This is an interesting report that helps to further the knowledge of fatigue and sleep in a rare disease population. Other authors have noted that OI is associated with pain & fatigue and interventions such as physiotherapy (Van Brussel, 2008) or bisphosphonates (Salehpour, 2010) may help the pain and/or fatigue. However, there is very little in the literature about OI and sleep and thus I welcome this large cross-sectional study which adds to our knowledge.

There are some minor issues with the study. Fatigue is a broad term and there are multiple factors contributing to fatigue. While the VAS fatigue scale is reasonable, the paper would have been much stronger by using an additional assessment for sleep such as the Epworth scale. Also, other sleep questionnaires would have been beneficial such as the Pittsburgh sleep quality questionnaire and the simple and short STOP-BANG questionnaire. Perhaps those could be included in any future research in this group.

Clearly, there is a higher incidence of sleep symptoms in the OI group as per Table 1. It may be useful to break out the 8 OI subjects who are diagnosed with sleep apnea (and on positive pressure ventilation) and see how their Table 1 scores compare to the the non-diagnosed group (presumably better on therapy).

Finally, it is unfortunate that the authors did not have access to the subjects medication list. Certainly part of the issue with poor sleep & fatigue is due to chronic pain and knowledge of pharmaceutical therapy would have been useful. Also, it would be useful in any future study to measure neck circumference since BMI in this population may be less accurate given height loss.

Overall, this is an interesting study that contributes to the knowledge of fatigue and sleep in this rare population.

Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.

Yes
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