Reviewer’s report


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Reviewer: Adriana Montaño

Reviewer’s report:

In this manuscript Chen et al have reported a case report of a patient with a p.G546S mutation in the COL2A1 gene. Although members of the family had the same genotype their phenotype was milder than that of the proband.

I think this case is interesting for the genetic community; however the authors did not produce any hypotheses that could explain this finding.

I have few concerns that need to be addressed:

1. The mutation in the COL2A1 gene was found by whole exome sequencing. The authors did not report presence of any polymorphisms found during sequencing. If the authors only looked at causative mutations it is very difficult to explain the difference in the patients' phenotype.

2. Authors should provide a hypothesis regarding the difference in phenotypes and how this could be explained (i.e. methylation, modifying factors, etc).

3. In table 1, please add the mutation found in each patient and also their demographics.

4. In figure 3b there was an alignment of different species showing the p.Gly546 residue; however this result was not mentioned or discussed in the text. Please provide the relevant information in the body of the paper.

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

Yes

Does the work include the necessary controls?
If not, please specify which controls are required in your comments to the authors.

Unable to assess

Are the conclusions drawn adequately supported by the data shown?
If not, please explain in your comments to the authors.

No

Are you able to assess any statistics in the manuscript or would you recommend an additional statistical review?
If an additional statistical review is recommended, please specify what aspects require further assessment in your comments to the editors.

Not relevant to this manuscript

Quality of written English
Please indicate the quality of language in the manuscript:

Needs some language corrections before being published

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