Reviewer’s report

Title: A nonsense PTH1R variant shows incomplete penetrance of Primary Failure of Eruption.

Version: 0 Date: 19 Oct 2019

Reviewer: Lorri Morford

Reviewer's report:

This is a well written case report presenting the findings of a 26-year old male patient diagnosed with Primary Failure of Eruption (PFE).

Only minor suggestions are made below:

1. In the abstract, because the study has been completed - the verb tense should be in the past tense. (i.e., The aim of this work "WAS" to describe a rare inheritance pattern of Primary Failure of Eruption (PFE)....

2. The authors mention the concept of reduced penetrance in their "Discussion and Conclusions" section. However, they do not provide any references that suggest that this has been observed in other families where one or more family member had been diagnosed with PFE. It would be helpful for them to either (a) provide reference(s) from the literature which demonstrate that reduced (or incomplete) penetrance has been observed in other family diagnosed with PFE, or (b) clearly indicate that this the first report of incomplete penetrance of PFE within a family.

3. Question: Did the son have any additional variations in his PTH1R gene copies (i.e., possibly a second less detrimental than the one reported at c.505G>T) which his mother did not inherit? I am simply trying to determine whether the son may have received a "double hit" - i.e., the c.505G>T variation in one PTH1R gene copy and a weaker variation in his second PTH1R gene copy which made haploinsufficiency of the PTH1R more of a problem for him (but not his mother).

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.

Yes

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

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:

Acceptable

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