Author’s response to reviews

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

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Author’s response to reviews:

A novel nonsense PTH1R variant shows incomplete penetrance of Primary Failure of Eruption: a case report. Grippaudo et al.

Dear Editor,

Many thanks for your invitation to provide a revised version of our manuscript. As requested here is a detailed list of the changes made and observations to the Reviewers’ comments.

Reviewer 1 suggested to investigate whether the patient’s mother had any bone-related conditions or osteoarthritis as previously reported by Frazier-Bowers et al. (Novel mutation associated with Primary Failure of Eruption and Osteoarthritis. J Dent Res 2014; 93:134-139). We were aware of their report and had already included it in the References, therefore we specifically asked the mother about her medical complaints and the only finding we could report was her relatively short stature (158 cm vs. 173 cm of her son and 171 cm of her husband).

Reviewer 2 made three observations:

1) The Reviewer asked to change the present to past tense at the beginning of the Abstract and we followed his/her suggestion. Pag.2 line 4
2) The Reviewer wondered if our report was the first to document incomplete penetrance of PFE and suggested to clearly state this in the manuscript. Actually, reduced penetrance has already been reported in some families including one reported by Frazier-Bowers et al. (J Dent Res 2014; 93:134-139), therefore we added half a sentence with the relevant reference in the Discussion and Conclusions section. Pag.5 line 50.

3) The Reviewer suggested that PFE might have been manifested only in the proband and not in his mother because he might have been carrying another “weak variant” on the paternal allele. This is an interesting suggestion but, as stated in the abstract and the Case presentation sections, we analyzed the entire PTH1R coding sequence and did not find any other “second hit” in the patient.

Again we wish to thank the Reviewers for their useful comments and suggestions and look forward to your Editorial decision.

Sincerely yours,

Cristina Grippaudo, D.D.S.