Author's response to reviews

**Title:** Variable expressivity of FGF3 mutations associated with deafness and LAMM syndrome

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**Author's response to reviews:** see over
Dear Dr. Norton,

Thank you for reviewing our manuscript entitled “Variable expressivity of FGF3 mutations associated with deafness and LAMM syndrome.”

We appreciate the helpful suggestion and comments, and provide a point by point response to them (see below). We are looking forward to hearing your decision regarding publication of our manuscript in BMC Medical Genetics.

Sincerely,

Byung Yoon Choi, MD, PhD
Communicating author

Responses to reviewers’ comments

Referee 1: Tom Walsh

1. Can authors comment on the possibility that the nucleotide alteration (c.283 C>T), underlying p.R95W, may affect splicing of the FGF3 exon in which it is located. For example, could the C>T mutation create a cryptic splice site mutation or perturb a potential exonic splice enhancer site that would lead to a ‘leaky’ splice mutation?

Thank you for your comment. In silico programs indicate that c.283 C>T is not predicted to affect splicing. We have added the following sentences on pages 9-10 of the Results section, which reads
“This variant (c.283C>T) has no predicted effect upon splicing using ESEfinder v3.0 (http://rulai.cshl.edu/cgi-bin/tools/ESE3/esefinder.cgi?process=home) and BDGP (http://www.fruitfly.org/se_tools/splice.html) programs. Therefore, it is unlikely that this variant creates a cryptic splice site or perturbs an exonic splice enhancer site that would lead to a leaky splice mutation.”

Referee 2: Frei Klemens

No comments