Reviewer's report

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

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Reviewer: Tom Walsh

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Riazuddin et al. report a genotype-phenotype study of FGF3 mutations and LAMM syndrome. They describe a novel FGF3 mutation and provide detailed clinical evaluation and molecular modeling of a previously described missense mutation (p.R95W).

Can the authors comment on the possibility that the nucleotide alteration (c.283C>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?

Level of interest: An article whose findings are important to those with closely related research interests

Quality of written English: Acceptable

Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:

I declare that I have no competing interests