Reviewers report

Title: NEXT-GENERATION SEQUENCING FOR IDENTIFYING A NOVEL/DE NOVO PATHOGENIC VARIANT IN A MEXICAN PATIENT WITH CYSTIC FIBROSIS

Version: 0 Date: 22 Jan 2019

Reviewer: Francesco Salvatore

Reviewers report:


This is a very important aspect in genetic counseling, and more so for diseases with high genetic heterogeneity such as Cystic Fibrosis. The case report is well written, and I agree for it to be published.

However, I would suggest some minor revisions to the authors:

1) the use of the NGS is presented to have a key role in this case report, but any references are directly related to the use of NGS in Cystic Fibrosis molecular diagnosis, although there are several examples in the literature (Ivanov M et al., 2018 BMC Med Genomics, Bergougnoix A et al., 2018 Clin Chem Lab Med, Lucarelli M et al., 2017 J Mol Diagn and so on). It would serve to reinforce the role of NGS technologies in CF.


A New Targeted CFTR Mutation Panel Based on Next-Generation Sequencing Technology. Lucarelli
2) The use of NGS is very important, but equally important is the functional characterization of the new variants identified. Also this concept, treated in part by the authors with the use of in silico predictions, should be treated also citing some examples from the literature (i.e, Hinzpeter A et al., 2017 Clin Case Rep, Terlizzi v et al, 2017 J Med Genet, Gianotti A et al, 2018 Front Pharmacol).

In vitro Methods for the Development and Analysis of Human Primary Airway Epithelia.
Gianotti A, Delpiano L, Caci E.


**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:

Needs some language corrections before being published

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