Reviewer’s report

Title: An Immunocompetent Patient with a Nonsense Mutation in NHEJ1 Gene

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Reviewer: Milena Cavic

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Title: "An Immunocompetent Patient with a Nonsense Mutation in NHEJ1 Gene"

COMMENTS TO THE AUTHORS

This study represents a rare case report of a homozygous carrier of a nonsense mutation in the NHEJ1 gene presenting with a general clinical immunocompetence at a tender-age. Clinical symptoms included failure to thrive, proportional microcephaly and an autoimmune hemolytic anemia, but the patient tolerated vaccination very well. The Manuscript is well written, the topic is interesting and the conclusions important for the clinical and scientific community. The experimental part of the study has been performed in an adequate manner and the obtained data analyzed using valid methods.

Minor comments:
1. Background, line 20: Please rephrase the sentence "One of the products of this process is DSB". DSB occur during V(D)J recombination, but it cannot be said that they represent the products of this process

2. Background, line 20: "One of the products of this process is DSB [3]". Reference 3 is not the original source of these data, please use a more appropriate reference

3. Case presentation, page 2, Line 31. Please introduce the abbreviation CMV with the corresponding full label of the virus

4. Case presentation, page 2, line 41. Please comment shortly on the values that are out of range in Table 1. and the possible causes/effects (CD8+ (%), CD19+ (%), CD14+ (%), CD56+ (NK cells), CD20+ (B cells marker), Plt (103/mm3), IgA (g/L))

5. Case presentation, page 2, line 57. Please add more methodological details on the WES methods here

6. Discussion and Conclusion, page 1, line 37: please rephrase "…variety of endogenously and exogenously..." to "…variety of endogenous and exogenous..."

7. Discussion and Conclusion, page 2, line 33: please rephrase "This mutation would lead to a
protein..." to "This mutation would lead to the production of a protein..."

8. Discussion and Conclusion, page 4, line 15: please rephrase "…initially diagnosed with AIHA, which responded..." to "…initially diagnosed with AIHA, but responded..."

9. The abbreviation list is incomplete, please add all the terms used in the manuscript

10. Table 1. Please add the Reference Values for CD45+ (%)

**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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