Reviewer’s report

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

Version: 0 Date: 18 Jan 2019

Reviewer: Fabio Concetti

Reviewer's report:

Overall, the manuscript "An Immunocompetent Patient with a Nonsense Mutation in NHEJ1 Gene" is well written and acceptable for publication after minor revisions.

The information from this case study is rather innovative and somehow surprising.

- Lines 14-41: it is not clear if clinical data shown were measured when the patient's age was 1 year, before or after this age. It must be stated because the "Case Presentation" section starts describing the patient when she was 3.5 years old and this is misleading. In Table 1 also, the age of the patient at the time the analyses were performed must be specified.

- Lines 33-36: it is stated that the stop-gain mutation described in this manuscript would lead to a protein lacking about one-third of its C-terminal. It would be appropriate to perform a western blot to confirm this fact in an incontrovertible manner, comparing the protein produced in the patient with appropriate healthy and pathological controls.

- TREC and KREC expression analysis from peripheral blood could be usefully added to this study to provide more information regarding the actual functionality of lymphocytes.

- Table 1: some values should be commented more extensively in the text, especially the WBC count showing leukopenia, the low level of CD19+ and the very high level of CD56+ cells and how can this framework be compatible with an immunocompetent phenotype as described in the paper's title? Absolute cell values should be written in the table and not only their percentages. How is DHR calculated? Is it a percentage? If so, how is possible to have 180? If not, why the reference value is a percentage whereas the patient's value is a number?

- It should be stated in the manuscript if there are special precautions (and/or drugs) taken to preserve patient's health (like a sort of low bacterial or germ-free environment) or if she lives a normal life.

Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.

No
Does the work include the necessary controls?
If not, please specify which controls are required in your comments to the authors.

Unable to assess

Are the conclusions drawn adequately supported by the data shown?
If not, please explain in your comments to the authors.

No

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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