Reviewer's report

Title: Identification of a novel nonsense mutation in SH2D1A gene from a patient with X-linked lymphoproliferative syndrom type 1: a case report

Version: 1 Date: 19 Jan 2018

Reviewer: Marta Olszewska

Reviewer's report:

Dear Authors,

your manuscript is interesting and can be published after some minor points:

1. English correction should be done by native speaker. In the manuscript, there are some strange grammar constructions that are not English. Additionally, some sentences are too long and need to be cut into 2-3 shorter ones, to give better flow of reading. Also, some commas are missing. Some suggestions are presented in an attachment.

2. Negative control should be explained in a paragraph with RT-PCR description.

3. Conclusions should be more directed into the fact, that you have found a novel mutation, rather (or simultaneously) that the apli-seq method role.

4. Some legend should be included into Figures' description.

Details you can find in reviewer's attachment.
**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.

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:

Needs some language corrections before being published

**Declaration of competing interests**
Please complete a declaration of competing interests, considering the following questions:

1. Have you in the past five years received reimbursements, fees, funding, or salary from an organisation that may in any way gain or lose financially from the publication of this manuscript, either now or in the future?

2. Do you hold any stocks or shares in an organisation that may in any way gain or lose financially from the publication of this manuscript, either now or in the future?

3. Do you hold or are you currently applying for any patents relating to the content of the manuscript?

4. Have you received reimbursements, fees, funding, or salary from an organization that holds or has applied for patents relating to the content of the manuscript?

5. Do you have any other financial competing interests?

6. Do you have any non-financial competing interests in relation to this paper?
If you can answer no to all of the above, write 'I declare that I have no competing interests' below. If your reply is yes to any, please give details below.

I declare that I have no competing interests.

I agree to the open peer review policy of the journal. I understand that my name will be included on my report to the authors and, if the manuscript is accepted for publication, my named report including any attachments I upload will be posted on the website along with the authors' responses. I agree for my report to be made available under an Open Access Creative Commons CC-BY license (http://creativecommons.org/licenses/by/4.0/). I understand that any comments which I do not wish to be included in my named report can be included as confidential comments to the editors, which will not be published.

I agree to the open peer review policy of the journal.