Reviewer’s report

Title: A Novel Mutation in SEPN1 Causing Rigid Spine Muscular Dystrophy 1: A Case Report

Version: 0 Date: 06 Oct 2018

Reviewer: Santasree Banerjee

Reviewer's report:

In this study, the authors performed a genetic molecular study to identify a Novel mutation in SEPN1 gene Associated with Rigid Spine Muscular Dystrophy 1.

Major Points:

1. In this study, please illustrate the clinical details in more comprehensive and detailed way. Please give the clinical details for the other family members.

2. In this study, please describe the methodology for identifying the mutation? Please give us the detailed process and quality control system for identifying the candidate variants.

3. The variant has been written according to the HGVS nomenclature rules. Please check and confirm it.

4. Please do a functional study of this identified mutation. It is a missense VUS, so functional characterization is required to confirm the pathogenicity. Please do a functional analysis of this mutation by in vitro or in vivo experiments and confirm how this mutation causes the disease.

5. There are some typos and grammar mistakes in the text and need some editorial improvement.

6. Please, delete irrelevant "general" phrases and sentences.

7. List up and explain all the abbreviation used in this manuscript (SCC, CHH, RJH).

8. Overall written English need to improve.

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.

I am able to assess the statistics

Quality of written English
Please indicate the quality of language in the manuscript:

Not suitable for publication unless extensively edited

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.
There is 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