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: Nanna Witting

Reviewer's report:

The study describes a single subject with a novel mutation in the SEPN1 gene.

The clinical phenotype is almost not described, only a very illustrative photo (fig 1) convinces the reader that this is indeed a case of rigid spine disease.

The majority of the text concerns general issues not specifically related to the subject. No effort is done in order to comment on whether this subject with a presumed patogenic novel SEPN1 variant differs from previously reported SEPN1 phenotypes.

Taken together no additional information is given than could be retracted from an mutation database.

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.

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:

Needs some language corrections before being published

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