**Reviewer's report**

**Title:** A novel missense mutation in the MYH7 gene causes an uncharacteristic phenotype of myosin storage myopathy: a case report

**Version:** 0  **Date:** 05 Dec 2018

**Reviewer:** Craig Kinnear

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The manuscript describes a case study of patient with a myosin storage myopathy where a novel disease-associated variant in the gene encoding the slow/cardiac beta myosin heavy chain (MYH7). The manuscript give an adequate description of the case and provides some evidence that the c.1370T>G variant in MYH7 is the disease-associated variants. I do have a few minor comments that should be addressed by the authors.

1) Please provide some more information about the way that the variants were filtered and prioritized following exome sequencing.

2) The variant is classified as likely pathogenic by the authors following results obtained from VarSome. Does this hold true if one applies the recommended guidelines from the American College of Medical Genetics and Genomics and the Association for Molecular Pathology (Genet Med. 2015 May ; 17(5): 405-424. doi:10.1038/gim.2015.30.)? Please use these guidelines to classify the variant.

3) I have noted that the necessary controls were not included. In order to confirm that this variant is disease-associated, one should be able to show that it segregates within the family. I do accept that this was not possible due to the lack of clinical information and genetic material available for the index case's relatives.

4) The manuscript contains a number of grammatical errors that needs to be addresses.
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.

No

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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