**Reviewer’s report**

**Title:** Novel phenotypic variant in the MYH7 spectrum due to a stop-loss mutation in the C-terminal region: a case report

**Version:** 0  **Date:** 31 Jan 2017

**Reviewer:** valeria novelli

**Reviewer’s report:**

The case report is very interesting but should be more clear in the description of the gene structure, on the phenotypes associated with variants in MYH7 gene and on the organization of the entire article.

I suggest to be more schematic in the Background: 1) Description of the phenotype 2) description of the gene and protein (figures of the structure could be added) 3) Clear and detailed description of similar variants reported in literature.

Methods are not reported. The electropherogram of the variant it is not clear. The two peaks of the variant are not specific. I suggest to repeat the sequence in Forward and Reverse (as double confirmation) and resubmit a new figure.

Details on the other two reported variants (rs#) and accession number (NM_) of the genes should be included.

I suggest to add the "Discussion" and summarize the "Conclusions".

The use of word "mutation" is not appropriate for variants without a clear attested pathogenicity.

English need to be revised.

**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:

Needs some language corrections before being published

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