Reviewer’s report

Title: A case report of recessive restrictive cardiomyopathy caused by a novel mutation in cardiac troponin I (TNNI3)

Version: 0 Date: 24 Jan 2019

Reviewer: Aldrin Gomes

Reviewer’s report:

This is a straightforward report of the discovery of a novel TNNI3 mutation: NM_000363.4:c.586G>C, p.(Asp196His) in twin sisters. Homozygous patients showed mainly a restrictive phenotype, but a homozygous brother showed a hypertrophic phenotype. Heterozygous family members were asymptomatic suggesting a recessive mode of inheritance.

The manuscript was well written and suggests that this region of troponin I may be a hotspot for RCM mutations.

Minor Concerns

Discussion should be expanded to include that this region may be a hotspot for HCM, as more RCMs have been found in this region than on any region in any other protein. A small paragraph on the likely effect of the Asp196His mutation on the function of the troponin I would be beneficial to the readers.

1A contains a slight misalignment of the vertical line form from I to II.

In figure 1 Legend. Please state unknown zygosity for twins.

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.

Not relevant to this manuscript

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

Acceptable

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