Reviewer’s report

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

Version: 0 Date: 15 Jan 2019

Reviewer: D. Iancu

Reviewer’s report:

Dear authors,

It was a pleasure to review your article "A case report of recessive restrictive cardiomyopathy caused by a novel mutation in cardiac troponin I (TNNI3)".

Genetic heterogeneity, incomplete penetrance and variable expression of cardiomyopathies have a negative impact on diagnosis yield and also on early identification of individuals at risk.

While the consanguinity, lack of symptoms in heterozygotes and presence of the disease in homozygotes only, can indeed support a recessive inheritance, this is more difficult to demonstrate for this condition, knowing the incomplete penetrance and phenotype variability. Therefore, in my opinion, there are a few elements that can be added to support the conclusion:

1) Given that the mutation meets the criteria to be considered damaging, it would be interesting to suggest a hypothesis as to why this mutation, compared to others located nearby and in a conserved region, is not also pathogenic in heterozygous individuals.

2) Related to this, it might be of interest to discuss the meaning of finding "Increased apical trabeculations" in I2. After analysing the electrocardiographic results for this individual, can you completely exclude a minor phenotype in this person? (please also see: PMID: 30279906).

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:

Acceptable

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