Author’s response to reviews

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

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Author’s response to reviews:

Dear Helena Kuivaniemi,

Thank you for your tremendous assessment/review.

Please see below our comments/answers for this round 3 of reply.

Regards,

Please, please carefully read your text. It still contains a number of errors. For example (there might be others, these are just examples of what I found with a quick search):

Agree!

We’ve carefully read our text and have found some minor errors. They have been fixed.

1) p. 3, line 14: change to "being overweight"

Accepted!
Change has been made as follow:

Page 3, line 12: …was mainly related to being overweight.

2) p. 6, line 3: change "variant mutation" to "variant"

Accepted!

Change has been made as follow:

Page 5, Line 26-27: … the MYH7 gene is a non-synonymous single nucleotide variant possibly linked to the clinical findings …

3) p. 6, line 11: "However, both mutations have been after filtering...” BAD sentence!

Accepted!

Change has been made as follow:

Page 6, lines 2-4: However, both mutations have been discarded after we filtered in only genes involved in neuromuscular functions that potentially cause clinical features of muscle myopathy observed in this patient (Additional file 2: Figure S1).

4) p.6, line 18: "result of analyse", maybe "results of analyses"....

Accepted!

Change has been made as follow:

Page 6, line 8: Results of analyses obtained from…

5) p. 6, line 20: do not use "verdict". That belongs to courtrooms, not scientific literature.

Accepted!

However, we just used the same employed on Varsome. However, to make it much more scientific literature rather than in courtrooms field, we’ve made changes as follow:

Page 6, lines 9-10: suggested a classification of Likely pathogenic for this variant,…

Page 6, line 14: …suggested the same classification of Likely pathogenic for this variant,…
6) Figure S1: define acronyms in the figure legend.

Accepted!

Acronyms added in the figure legend as follow:

AR: autosomal recessive; AD: autosomal dominant; AQ: alignment quality; ExAC: The Exome Aggregation Consortium; HGMD: The Human Gene Mutation Database; gnomAD: The Genome Aggregation Database; NHLBI ESP: The National Heart, Lung, and Blood Institute Exome Sequencing Project

7) Figure S1: "inclusion bodies" has a typo

Accepted!

Correction has been made.

8) Table S3: Which score was used to determine "significance"?

Significances have been generated by our in-silico prediction tool using the overall score from SIFT, PolyPhen2, Grantham and PhyloP. For better relevance of the information we publish, we decided to apply the ACMG-AMP updated criteria. Accordingly, changes have been made directly in the column for significance and a note has been added in the footnote of the table (see track changes).