Author’s response to reviews

Title: A mutation in the filamin c gene causes myofibrillar myopathy with lower motor neuron syndrome: a case report

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Author’s response to reviews:

Dear Editors,

Thank you for accepting our manuscript entitled "A mutation in the filamin c gene causes myofibrillar myopathy with lower motor neuron syndrome: a case report" (NURL-D-18-00917R1).

The corrections in the paper and the responds to the Editor's comments are as following:

1. Responds to the Editor’s comments: the current submission contains some textual overlap with other previously published works, in particular: (1) Filamin C Truncation Mutations Are Associated With Arrhythmogenic Dilated Cardiomyopathy and Changes in the Cell–Cell Adhesion Structures. (2) Pathophysiology of protein aggregation and extended phenotyping in filaminopathy. (3) Handbook of Clinical Neurology, Chapter 140 - Myofibrillar myopathies
We are very sorry for our negligence of containing textual overlap with other previously published words. We have changed our expression as below:

1. Line 65-68, Page 3, “Filamin C (FLNC) is an actin cross-linking protein and also one of the largest Z-disk proteins in cardiac and skeletal muscle. Its main function is to connect muscle cells to the extracellular matrix and be involved in related signaling pathways.” was corrected.

2. Line 68-71, Page 3, “The filamin C-related myopathy was first described in 2005. The patients from a large German family with the nonsense mutation (c.G8130A, p.W2710X) in the FLNC immunoglobulin(Ig) like domain 7 had predominant limb-girdle muscle weakness.” was corrected.

3. Line 161-166, Page 7, “MFMs are rare, hereditary and progressive muscle disorders with distinctive clinically and genetically heterogeneous.[7] They are defined by the presence of myofibrillar disorganization commencing at the Z-disk, an accumulation of myofibrillar degradation products, and the ectopic expression of multiple proteins. MFM subtypes are designated according to the affected protein, such as desminopathy, aB-crystallinopathy, Bag3opathy, or filaminopathy.” was corrected.

2. Responds to the Editor’s comments: Please clarify why you have changed your ethics approval statement to now read 'Not applicable'.

Last view letter that the Editor sent us on 13 May included this comment, “Please clarify why ethics approval was obtained for this Case Report. Generally, and according to most countries' national regulations, case reports do not require formal ethics approval unless they are reporting the experimental use of a novel procedure or tool. If you did use a new procedure or tool on the patient, please make this clear in the manuscript and provide a clear justification for why the new procedure or tool was deemed more appropriate than usual clinical practice to meet the patient’s clinical needs. If this was not the case, simply state "Not applicable" in the Ethics section.” Since we did not use a new procedure or tool on the patient, so we just simply state "Not applicable" in the Ethics section, and deleted “ The study protocol was approved by the Ethics Committee of Peking University Shenzhen Hospital in compliance with the Declaration of Helsinki.” But in the case report, we tested the DNA of 100 healthy unrelated controls to identify that the mutation (c.7123G>A, p.V2375I) is probably a new pathogenical mutation. And this study was approved by the Ethics Committee of Peking University Shenzhen Hospital.
3. Responds to the Editor’s comments: Please include a statement in the Authors' contributions section to the effect that all authors have read and approved the manuscript, and ensure that this is the case.

Line 301-302, Page 13, “All authors have read and approved the final manuscript, and ensure that this is the case.” was added.

4. Responds to the Editor’s comments: Thank you for providing your completed CARE checklist as an additional file. As this is no longer required at this stage please remove this when uploading the revised version of your manuscript.

We will remove the CARE checklist as an additional file when uploading the revised version of our manuscript.

5. Responds to the Editor’s comments: Please add a section "Additional files" (after the References/Figure legends).

Line 388-393, Page 17, “Additional file 1 Nerve conduction studies and electromyography results. The nerve conduction velocity revealed severe reduction in CMAP amplitudes and motor conduction velocities in the left ulnar nerve(A), while the sensory conduction was normal(B). Motor unit action potentials of increased amplitude and duration was observed in the EMG, including the right sternocleidomastoid(C), left biceps brachii(D) and left vastus medialis(E).” was added. And all figures, tables and supplementary/additional files are cited within the text (Line 114 and 118, Page 5).

6. Responds to the Editor’s comments: At this stage, please upload your manuscript as a single, final, clean version that does not contain any tracked changes, comments, highlights, strikethroughs or text in different colours. All relevant tables/figures/additional files should also be clean versions. Figures (and additional files) should remain uploaded as separate files. Please ensure that all figures, tables and additional/supplementary files are cited within the text.

We are sure that our manuscript is a single, final, clean version that does not contain any tracked changes, comments, highlights, strikethroughs or text in different colours. All relevant tables/figures/additional files is clean versions. Figures (and additional files) will be uploaded as separate files. All figures, tables and additional/supplementary files are cited within the text.
We tried our best to improve the manuscript and made some changes in the manuscript. These changes will not influence the content and framework of the paper. We appreciate for Editors’ warm work earnestly, and hope that the correction will meet with approval.

Once again, thank you very much for your comments and suggestions

Best wishes,

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