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 and Reviewers,

Thank you for your letter and for the reviewers’ comments concerning 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). Those comments are all valuable and very helpful for revising and improving our paper, as well as the important guiding significance to our researches. We have studied comments carefully and have made correction which we hope meet with approval. Revised portion are marked in red in the paper.

The main corrections in the paper and the responds to the Editor and reviewer’s comments are as following:

1. Responds to the Editor’s comments: Please clarify why ethics approval was obtained for this Case Report.
Since we did not use a new procedure or tool on the patient, so we just simply state "Not applicable" in the Ethics section. Line 293-294, “The study protocol was approved by the Ethics Committee of Peking University Shenzhen Hospital in compliance with the Declaration of Helsinki.” was deleted.

2. Responds to the reviewer’s comments:

Reviewer #1:

1. Response to comment: Did the authors confirm the FLNC variant by Sanger sequence?
   
   Line 151-153, “Confirmation of the variant was undertaken by Sanger sequencing using an ABI 3730XL DNA Sequencer (Applied Biosystems, Thermo Fisher Scientific, USA),” was added.

2. Response to comment: It is better to add the information about the family members including genetic examinations, if possible.

   Since the patient had no immediate family members and loose contact with other family members, further co-segregation analyses among the family cannot be conducted. This was also added in the Line 159-161.

Reviewer #2:

1. Response to comment: There are no details of the electrophysiology evaluation: how slow were the motor conduction? was sensory conduction tested? Were there fasciculations in the muscles tested (especially those that showed clinical fasciculation)? which muscles were examined in lower and upper limbs? Were other nerves’ Fawaves elicited and at what latency?

   It was for the sake of brevity we did not show the details of the electrophysiology evaluation, but we will add the results of the NCV and EMG in the supplementary documents. The motor never conduction velocity (MCV) of left ulnar n. and radial n. was 40m/s. The MCV of the left median nerve cannot be stimulated. The sensory nerve conduction was normal in all these nerves. There was no fasciculations in the muscles tested, but spontaneous activity (positive sharp waves) was recorded. We tested the left bicep and triceps brachial muscle, right tibialis anterior muscle, left vastus medialis muscle, right sternocleidomastoid muscle, and paraspinal muscle. We just elicited the F waves of the ulnar n.
2. Response to comment: However one would like to know if their NGS excluded all motor neuron disease gene defects?

We have recheck the result of NGS, fifty-six motor neuron disease gene were normal, such as ANXA11, ASCC1, ATP7A, SOD, SMN1 and so on.

3. Response to comment: Were there family members of the patient available for testing?

Since the patient had no immediate family members and loose contact with other family members, further co-segregation analyses among the family cannot be conducted. This was also added in the Line 159-161.

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. And here we marked the changes in red in revised paper. We appreciate for Editors/Reviewers’ warm work earnestly, and hope that the correction will meet with approval.

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

Yours sincerely,

Wu Jun