Author’s response to reviews

Title: Muscular involvement and tendon contracture in limb-girdle muscular dystrophy 2Y: a mild adult phenotype and literature review.

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Editor Comments:
The quality of the English used throughout your manuscript does not currently meet our requirements, as there are several incorrect sentence constructions and grammatical errors throughout obscuring the message the authors want to convey. We recommend that you ask a native English speaking colleague to help you copy-edit the paper. If this is not possible, you may need to use a professional language editing service. Use of an editing service is neither a requirement nor a guarantee of acceptance for publication.

We operate a transparent peer review process for this journal where reviewer reports are published with the article but the reviewers are not named (unless they opt in to include their name).

A : The revised manuscript has been reviewed and improved by AJE(American Journal Experts).

Reviewer reports:
Reviewer 1: One main point is: the text should be fully improved by a native speaker.

A : The revised manuscript has been reviewed and improved by AJE(American Journal Experts).
The authors describe an additional patient suffering from LGMD2Y. "Skeletal dysmorphia" in the title is misleading, a contracture is not a skeletal dysmorphia.

A: We have changed "Skeletal dysmorphia" into “tendon contracture”.

In the clinical feature part, the heart examination of the patient should be mentioned.

A: Unfortunately, we did not have detailed heart examination of patients at diagnosis and patients rejected further examination after diagnosis.

Results of electromyography should be described in more detail.

A: We had expanded the description of EMG result(Result section, line 80).

In the shown histological photos and EM photos the observed abnormalities (sometimes described rather diffusely) should be pointed out by arrows.

A: We had modified our figure according to your suggestion.

Please note the use of "variant" and "mutation".

A: We had corrected this mistake(Result section, line 111)

The sequencing procedure and following data analysis should be explained in more detail.

A: We had added more details of sequencing procedure and data analysis in this part (Result, section, line 102)

In quoted reference 5, part of the discussion (table 1!) is already anticipated. The authors should restrict it to the "real" muscle cases. The enrolvement of one or two isoforms of LAP1 and the consequences are already intensively discussed in reference 5, there is nothing new to add. Figure 4b is not really necessary. Nevertheless he mild phenotype of the newly described patient is quite interesting.

A: The discussion of present case was inspired a lot by reference 5. We had modified this part(Discussion section, line 149) and change Fiugre.4 as your suggestion.

Reviewer 2: In the manuscript by Feng et al., the authors describe the phenotype of a case with LGMD2Y who has a novel. This is an important paper because currently TOR1AIP1 has only been reported in a limited number of individuals and consequently is not even recognized as a LGMDR under the new nomenclature (Straub et al., 2018). However, to be truly useful, additional information about the phenotype is required.
1. I'm not sure that this patient had skeletal dysmorphia. It appears that he just had Achilles tendon contracture which is common in a variety of muscular dystrophies. I would remove skeletal dysmorphia from the title of the manuscript and text.

A: We had removed ‘skeletal dysmorphia’ from the title.

2. LGMD2Y is also characterized with spine rigidity. Was this a feature of the current case?

A: Neither spine rigidity nor scoliosis was found in current case.

3. What is meant by perimysium and endomysium were "enlarged"? Do the authors mean that there was increased fibrosis? The histopathology are not very suggestive of that.

A: We meant that the fibrosis in perimysium and endomysium was increased and we had modified this description (Result section, line 86). Since the present histopathology image was not suggestive enough, we had uploaded another HE image with more apparent fibrosis and inflammation.

4. Line 100, the authors mean "homozygous" not "homogenous"

A: We had corrected this mistake.(Result section, line 106)

5. Was the mother heterozygous for the same mutation?

A: The patient's mother was heterozygous for the same c.98dupC mutation.

6. Please describe the patient's cardiac function (by echo or cMRI).

7. Please describe the patient's pulmonary function (by spirometry).

A: Unfortunately, we did not have detailed examination of heart and lung at diagnosis and patients rejected further examination after diagnosis.