Reviewer’s report

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

Version: 0 Date: 28 May 2020

Reviewer's report:

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.
2. LGMD2Y is also characterized with spine rigidity. Was this a feature of the current case?
3. What is meant by perimysium and endomysium wer "enlarged"? Do the authors mean that there was increased fibrosis? The histopathology are not very suggestive of that.
4. Line 100, the authors mean "homozygous" not "homogenous"
5. Was the mother heterozygous for the same mutation?
6. Please describe the patient's cardiac function (by echo or cMRI).
7. Please describe the patient's pulmonary function (by spirometry).

Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.

Yes

Does the work include the necessary controls?
If not, please specify which controls are required in your comments to the authors.

Yes

Are the conclusions drawn adequately supported by the data shown?
If not, please explain in your comments to the authors.

Yes

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:

Needs some language corrections before being published

**Declaration of competing interests**
Please complete a declaration of competing interests, considering the following questions:

1. Have you in the past five years received reimbursements, fees, funding, or salary from an organisation that may in any way gain or lose financially from the publication of this manuscript, either now or in the future?

2. Do you hold any stocks or shares in an organisation that may in any way gain or lose financially from the publication of this manuscript, either now or in the future?

3. Do you hold or are you currently applying for any patents relating to the content of the manuscript?

4. Have you received reimbursements, fees, funding, or salary from an organization that holds or has applied for patents relating to the content of the manuscript?

5. Do you have any other financial competing interests?

6. Do you have any non-financial competing interests in relation to this paper?

If you can answer no to all of the above, write 'I declare that I have no competing interests' below. If your reply is yes to any, please give details below.

I declare that I have no competing interests

**Reviewer Publication Consent.** I agree for my report to be made available under an Open Access Creative Commons CC-BY License ([http://creativecommons.org/licenses/by/4.0](http://creativecommons.org/licenses/by/4.0)) if this manuscript is accepted for publication. Any comments that I do not wish to be included in the published report have been included as confidential comments to the editor, which will not be published. If you are not happy for us to publish this report, please contact the editorial office before completing the review. If you wish, you can include your name in your published report. Please note you must decide whether to include your name at the start of the process and confirm this decision whenever prompted. If you change your decision later, we will not be able to publish your name.

I agree to the terms of the CC-BY 4.0 license; please do not publish my name with my report. (default)