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:** 18 May 2020

**Reviewer's report:**

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

The authors describe an additional patient suffering from LGMD2Y. "Skeletal dysmorphia" in the title is misleading, a contracture is not a skeletal dysmorphia. In the clinical feature part, the heart examination of the patient should be mentioned. Results of electromyography should be described in more detail. In the shown histological photos and EM photos the observed abnormalities (sometimes described rather diffusely) should be pointed out by arrows. Please note the use of "variant" and "mutation". The sequencing procedure and following data analysis should be explained in more detail. In quoted reference 5, part of the discussion (table 1!) is already anticipated. The authors should restrict it to the "real" muscle cases. The involvement 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 the mild phenotype of the newly described patient is quite interesting.

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

No

**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:

Not suitable for publication unless extensively edited

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