Author's response to reviews

Title: Late-onset myopathy of the posterior calf muscles mimicking Miyoshi Myopathy but not related to a dysferlin mutation: Case report

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Author's response to reviews:

Re: Late-onset myopathy of the posterior calf muscles mimicking Miyoshi Myopathy but not related to a dysferlin mutation: Case report

Dear Professor Michael Kidd, dear editorial team,

We resubmit this paper with additional data to address the concerns of the referees and with the revisions that were proposed by the referees.

We thank you for your careful editorial attention, and we thank the reviewers for their engaged and detailed comments. We hope, you will agree that the enclosed revised manuscript meets your high standard for relevance and novelty.

Would the editorial team and the referees accept if we changed the title into?:

A novel anoctamin 5 mutation mimicking Miyoshi Myopathy: a case report

Point to point response:

Referee 1 Simone Spuler

We thank the reviewer for her important comments. As proposed by the reviewer (and the editorial board), we included the ethnic background of the patient and in additions added a photograph of the patient's legs (Fig. 1A). Furthermore, we stressed –as proposed by the reviewer- that dysferlin immunoblotting is more specific than immunostaining. The discrepancy of immunostaining and immunoblotting is now documented in figure 2. As proposed by the reviewer, we removed the normal dysferlin staining (control).

Minor: Affiliation 4 was assigned to the respective author.

Referee 2 Rumaisa Bashir

We thank the reviewer for his important comments. We have added details of the mutation including the involved exon/intron. We included the sequence data (sequencing electrospherogram) in Fig. 2 as proposed by the reviewer. We stressed in the abstract and in the case presentation section that the detected mutation is novel. The mutation was not previously described in literature nor in
the databases (minor point 3). We also hypothesize which possible effect the mutation could have on the protein. Furthermore, we added - to the conclusion section - that the mutation is associated with reduced sarcolemmal dysferlin and cytoplasmic dysferlin accumulation (minor point 6).

Minor 1, 2, 5: spelling and reference errors have been corrected

We would like to thank the referees for their careful and thorough critique of our paper. We have attempted to address the issues raised by each of the referees and in doing so, we believe the paper is markedly improved. We hope that you will reconsider the revised manuscript for publication in the Journal of medical case reports.

Yours sincerely

Clemens Neusch