Reviewer's report

Title: COL6A1 Mutation Leading to Bethlem Myopathy with Recurrent Hematuria: A Case Report

Version: 1 Date: 12 Aug 2018

Reviewer: Edvard Ehler

Reviewer's report:

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An interesting case report, concentrated on genetics and biochemistry.

My comments:

1. the clinical findings are not enough described - e.g., face, tongue, shoulders, swallowing, handgrip, muscle contractions, etc
2. EMG - polyspike and polyphasic MUP - but no duration and amplitude are added

Unfortunately, renal biopsy was not performed.

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?
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Yes

Are the conclusions drawn adequately supported by the data shown?
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Not relevant to this manuscript

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