Reviewer’s report

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

Version: 1 Date: 02 Dec 2018

Reviewer: Anne Güttsches

Reviewer's report:

The authors describe a case of a 14-year old boy with bethlem myopathy and recurrent hematuria. In the discussion, the possible connection between bethlem myopathy and hematuria are described, including the involvement of different collagens. This is an interesting idea of clinical relevance. I have some remarks:

On page 3, l 45 the authors describe a "treatment". Which treatment did the patient receive?

Did the authors exclude other reasons for hematuria? Was renal function normal at discharge? As a renal biopsy could not be taken, I doubt that a kidney disease could be excluded properly.

Did the authors perform a collagen VI- stain (i.e. by immunofluorescence)? It would be interesting to know, if the mutation in the COL6A1- gene leads to a loss or reduction of collagen- VI fibers in skeletal muscle tissue.

Concerning language:

Throughout the manuscript, the language has to be improved. I guess the reader will know, what the authors want to say, and is able to understand it. However, especially the discussion needs revision, as some parts are not clear.

One example: "The missense mutation in COL6A1, c.877G>A, in the conserved Gly-X-Y motif in the triple helical domain, altered structure of the α1 chain and then the structure of collagen VI." (p.4, ll14-17)

To accept the manuscript for publication, the manuscript, especially the "discussion and conclusions"- part, has to be edited.
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.

Unable to assess

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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