Reviewer's report

Title: Prune Belly Syndrome in Surviving Males can be caused by Hemizygous Missense Mutations in the X-linked Filamin A Gene

Version: 0 Date: 31 Jul 2019

Reviewer: Lyuba Miteva

Reviewer's report:

Summary

The manuscript titled: "Prune Belly Syndrome in Surviving Males can be caused by Hemizygous Missense Mutations in the X-linked Filamin A Gene" submitted to the BMC Medical Genetics with manuscript number: MGTC-D-19-00144 have reported three new point mutations in Fulamin A gene ((c.4952 C&T (p.A1448V), c.6727C&T (p.C2160R), c.5966 G&A (p.G2236E)), located in X-chromosomes among 4 cases of prune belly syndrome (PBS). Since the etiology of PBS remains largely unknown I find the paper as valuable, in the focus of the journal, with scientific potential, and well written. In addition, I highly appreciate that authors have shown the functional significance of the mutations in a view of the PBS etiology.

However, some recommendations I should mention.

Recommendations:

METHODS:

1. Since X-chromosome inactivation results are given in Supplementary Figure 1 I recommend the subheadings X-chromosome inactivation to give briefly in the figure's legends and erase From Methods section.

2. The subheading "FLNA expression studies" should be changed as gene expression studies, with respect to ITGB1 expression analysis. How many samples /biological repeats/ were included in FLNA expression studies?

3. Please, explain why you perform gene expression assays across normal adult human RNA samples and Immunohistochemistry on bladder tissue isolated from normal pediatric PBS-unaffected individuals? Don't you expect the age-related or disease-related differences? Do you have information about FLNA expression across PBS cases?
RESULTS:
4. Please, arrange the results section to achieve logical and easier reading. Figures and Tables should be given in consequent order.
5. Please, limit the discussion elements in the results.

DISCUSSION:
6. The discussion should be shortened and focused.
7. The limitations of the study should be included.
8. A reference list should be shortened and in the order that they occur in the text
9. List of abbreviations should be in alphabetical order.
* Minor point: Use double line spacing in preparing main manuscript text

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.
I am able to assess the statistics
Quality of written English
Please indicate the quality of language in the manuscript:

Acceptable

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