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: 14 Aug 2019

Reviewer: Kentaro Mizuno

Reviewer's report:

General comments:

The genetic cause of Prune Belly Syndrome (PBS) remains unknown, and authors focused on candidate genes fit an X-linked recessive mode of inheritance. In this study, authors have identified three hemizygous mutations (p.Cys2160Arg, p.Ala1448Val, p.Gly2236Glu) in FLNA gene in one multiplex kindred and two sporadic PBS males using whole exome sequencing. Beside, authors have also shown distribution of FLNA protein and disruption of binding to integrin. This is the first evidence for an X-linked cause of PBS in multiple unrelated individuals and expands the phenotypic spectrum associated with FLNA in males surviving even into adulthood.

Specific comments:

This paper is well-written, structured article. Although PBS is rare disease, authors enrolled individuals with PBS and their family. Authors' efforts brought the excellent result, and this article would be one of the most important in the research field of PBS hereafter.

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
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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
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