Reviewer’s report

Title: Case report: two novel VPS13B mutations in a Chinese family with Cohen syndrome and hyperlinear palms

Version: 0 Date: 31 Jul 2019

Reviewer: Matthew Butchbach

Reviewer's report:

This case report describes a novel compound heterozygous set of mutations in the VPS13B gene that is linked to Cohen syndrome. Overall, this a straightforward report of novel mutations associated with this rare disease. There are some points which must be addressed:

1. The VPS13B p.K3282X mutation is a nonsense mutation, not a missense mutation. Nonsense mutations insert a premature stop codon within the reading frame while missense mutations simply can the amino acid sequence.

2. The rationale for using exome sequencing for this family instead of more direct targeting sequencing of the VPS13B gene is not provided. Since most, if not all, Cohen syndrome cases are associated with mutations in VPS13B, would it not make better sense to sequence this gene first instead of using the more expensive, more labor intensive and more data intensive exome sequencing approach?

3. Figures 1 and 2 should be merged into a single figure.

4. The authors states, on more than one occasion, that "China is the most populated country in the world." What is the relevance of this statement to this manuscript?

5. The quality of the written English in this manuscript is poor. The authors should consider soliciting editorial assistance.
Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.

Unable to assess

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.

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Please indicate the quality of language in the manuscript:

Not suitable for publication unless extensively edited

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