Reviewer’s report

Title: First case report of Cohen Syndrome in the Tunisian population caused by VPS13B mutations

Version: 1 Date: 07 Aug 2017

Reviewer: Éliane Chouery Chouery

Reviewer’s report:

I highly appreciated the request to review this paper proposal. I am really sorry for the delay. I have carefully reviewed it and you will find below some comments.

1- in abstract, case presentation: authors should add the pigmentary retinopathy found in the girl; a clinical sign important in Cohen syndrome.

2- in the paper, case presentation: authors should add testing of neutropenia, it is important to know if these patients present neutropenia or not even after the WES was performed. Neutropenia is a clinical sign important in Cohen syndrome, if these patients after confirmation at the molecular level do not present neutropenia, it will add value to the paper.

3- in genetic testing: authors should add briefly the technology used for exome sequencing, which Platform? which Bioinformatics Tools were used? how many variants were detected? how did they filter variants to obtain these excellent results?

BEST REGARDS AND GOOD LUCK

Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.

No

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.

Not relevant to this manuscript

**Quality of written English**
Please indicate the quality of language in the manuscript:

Needs some language corrections before being published

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