Reviewer’s report

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

Version: 1 Date: 21 Aug 2017

Reviewer: Lionel Van Maldergem

Reviewer’s report:

The paper by Rejeb et al describes a sibship with two children presenting a paucisymptomatic form of Cohen syndrome. Atypical cases are not exceptional in CS and it usually does not make sense to publish case reports of a well-known syndrome already reported and delineated so many times in medical literature. However, the current report draws attention to redefinition of clinical spectrum of a specific entity by means of systematic whole exome sequencing. In this regard, the paper deserves consideration. Language is poor and would require checking by a native English speaker. The authors made the effort of reviewing literature but the paper describing glycosylation defects (Hum Mol Genet) is lacking. I am wondering whether a letter format would not be more appropriate. Some minor comments:

L22 delete « in »

L26 « hypotonia »

P3L14 delete gene

P4L6 "The parents gave their consent "should take place elsewhere

Two patients

From a non-consanguineous

"At least" delete

L33 When reevaluated at 12 y, growth retardation and progressive microcephaly were noted

P5L1 Her brother

L52 was validated

P6L1 were of biparental origin

L19 delete extremely
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:

Needs some language corrections before being published

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