Reviewer’s report

Title: Broadening the phenotype of the TWNK gene associated Perrault syndrome

Version: 0 Date: 26 Sep 2019

Reviewer: Imen chakchouk

Reviewer's report:

Bálint Fekete et al presented "Broadening the phenotype of the TWNK gene associated Perrault syndrome". In this study, they are describing new phenotypes ascertained in patients with TWNK-dependent Perrault syndrome. The authors reported medulla oblongata and cervical spinal cord atrophy, moderate cortical atrophy associated with Perrault syndrome.

Genetic investigation revealed multiple mtDNA deletion and compound heterozygous mutations of the TWNK gene (c.1196 A&gt;G, c.1358 G&gt;A).

The manuscript is very well written, very clear and organized. I just have few remarks:

- Author should include the Variant frequency from GnomAD as an extra proof of the pathogenicity of the variants beside the bioinformatic predictions. Only one c.1196 A&gt;G allele has been reported in east Asian population. c.1358 G&gt;A was reported once in the European non-Finnish.

- Author should also add the ethnicity of the patient previously reported that carried the variant c.1196 A&gt;G.

- I also have one simple question, why are you still using hg18? (Page5, line13)

- Add labels to the figures (A, B, C and D)
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.

Not relevant to this manuscript

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

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