Reviewer's report

Title: Novel likely pathogenic variants in TMEM126A identified in non-syndromic autosomal recessive optic atrophy: two case reports

Version: 0 Date: 14 Nov 2018

Reviewer: Dominique Bonneau

Reviewer's report:

The manuscript sent by Kloth et al. to the Journal reports the finding of novel pathogenic or likely pathogenic variants in TMEM126A in individuals from unrelated families affected with non-syndromic autosomal recessive optic atrophy (AROA).

This report is interesting because little is known about non-syndromic AROA probably due to the rarity of the disease. In addition, only one recurrent nonsense variant in TMEM126A has been reported so far in non-syndromic AROA in individuals all originating from Algerian Magrheb with a founder effect.

This is therefore, to my knowledge, the first report of additional pathogenic variants in TMEM126A, unequivocally confirming the involvement of this gene in non-syndromic AROA.

Pathogenicity of both variants is convincing even for the second missense one.

Minor comments:

- Please change the term "mutation" to "pathogenic variant" or "likely pathogenic variant" in the text and in the title as well.

- Please prefer the term "affected individuals" to "patients"

- Put TMEM126A in Italic type when dealing with the gene throughout the manuscript and abstract.
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:

Acceptable

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