Reviewer’s report

Title: A new genetic variant of hereditary Apolipoprotein A-I amyloidosis: a case-report followed by discussion of diagnostic challenges and therapeutic options

Version: 0 Date: 29 Nov 2018

Reviewer: Vassiliki Iconomidou

Reviewer's report:

This manuscript by Moutafi et al. reports on an APOA1 mutation, associated with hereditary ApoAI amyloidosis with liver and gonadal involvement. The authors use a range of appropriate techniques, including baseline lab tests and liver biopsy to characterize the fibrillar typing, whereas Gene sequencing and mutational analysis is used to unravel the APOA1 specific mutation. Both experimental and clinical results are clear and well-presented. Overall, this study emphasizes the need of amyloid typing in the diagnostic approach and would be of great interest to the BMC Medical Genetics readership.

I only have few minor comments:

- This Case study follows the CARE report. CARE guidelines should be suitably cited (for example: Gagnier J, Kienle G, Altman DG, Moher D, Sox H, Riley DS, and the CARE group. The CARE guidelines: consensus-based clinical case report guideline development. Journal of Clinical Epidemiology;67(1),46-51.)

- Page 10. "The amyloidogenicity of these mutations and the variability in the clinical presentation provides a complex model of interactions between the produced amyloid fibrils and the involved tissues" Authors are encouraged to elaborate on this statement by introducing a scheme of this hypothetical model.

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:

Acceptable

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