Reviewer’s report

Title: Whole Exome Sequencing Identified a Novel Truncation Mutation in the NHS Gene Associated with Nance-Horan Syndrome

Version: 0 Date: 03 Aug 2018

Reviewer: John Rosendahl Østergaard

Reviewer's report:

The authors describe a new mutation in the NHS-gene and discuss genotype-phenotype association in Nance-Horan syndrome contra X-linked Cataract. However, I do not find the conclusion: the study provided a novel insight into the understanding of genotype-phenotype association in NHS, sufficiently proven. In many aspects, the cases may not be representative and/or are not sufficiently described. For instance; as concern the teeth, a very important marker for the disease, I miss a description of the molars in all cases. Further, the affected two patients do not show any kind of mental disabilities, they have no nystagmus and were operated at an older age. In addition, the heterozygous females had no posterior suture or posterior stellate cataracts. Thus, the family history show a family that is not severely affected, as also stated by the author. Such cases should be used with more humility as done in the present paper when discussing a possible differences between the NHS disease and the X-linked cataract, especially as the dental characteristics are inadequately described.

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.

No

Are the conclusions drawn adequately supported by the data shown?
If not, please explain in your comments to the authors.

No

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:

Not suitable for publication unless extensively edited

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