Reviewer's report

Title: Segregation analysis in families with Charcot-Marie-Tooth disease allows reclassification of putative disease causing mutations.

Version: 2 Date: 26 November 2013

Reviewer: Feng Yue

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In the manuscript, the authors tried to use the family information to help evaluating genetic variants of unknown clinical significance for the CMT patients. The approach is not novel but of practical meanings. The work done here is solid.

Comments:

I found it interesting that 14.6% of the affected relatives tested as mutation negative. It will be great if the authors can hypothesize or explain why.

Minor:

1. how is reclassification defined?
2. how is CMT3 defined?
3. "Regarding the mutant gene identified in the index patients, the proportions of request for the testing of relatives were 23 % for PMP22 dup-, 60 % for GJB1, 64 % for MPZ and 42 % for MFN2." The number doesn't add up, it seems some patients carry mutliple mutations. Can auditors draw a pie chart?

Level of interest: An article of limited interest

Quality of written English: Acceptable

Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:

I declare that I have no competing interests.