Reviewer's report

Title: POLG1 R722H mutation associated with multiple mtDNA deletions and a neurological phenotype

Version: 3 Date: 24 February 2010

Reviewer: Michio Hirano

Reviewer's report:

Komulainen and colleagues have addressed criticism raised in the prior review. The additional data support pathogenicity of the POLG1 p.R722H mutation in homozygosity in a family with late-onset disease (mainly hearing loss and dementia with PEO in proband) and in more typically young-onset disease in compound heterozygosity with the p.W748S mutation.

Major Compulsory Revisions
None.

Minor Essential Revisions
None.

Level of interest: An article of importance in its field

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