Reviewer’s report

Title: A novel mitochondrial DNA mutation associated with hypertension in C4329A mutation between tRNA(Ile) and tRNAGln genes

Version: 4 Date: 28 May 2014

Reviewer: Frederic Tort

Reviewer’s report:

The paper by Liu et al. reported a new mutation in the mitochondrial DNA which is associated with hypertension in a Chinese family. Clinical follow-up of this family strongly suggested the possibility of maternal inheritance of hypertension. The authors screened the whole mitochondrial genome in individuals of this family in the search of mutations in the mtDNA that could explain these observations.

The study is, in general, well designed and the observations reported in the manuscript are of interest since the authors expand the spectrum of genetic alterations associated to hypertension.

The manuscript can be accepted with some minor and discretionary suggestions.

Minor essential Revisions:

1- Abbreviations: Some abbreviations used in the manuscript should be reviewed.
   - In page 3 (line3): CVD, cardiovascular disease.
   - In page 4 (line 11): cardiovascular disease could be abbreviated (CVD).
   - Page 9-10: in the “List of Abbreviations” section CVD is missing.

2- The reference list should be checked, since in the background section the first two references are annotated as (6) and (5) (page 3, first paragraph).

3- In-text figure citations:
   - I suggest that Figure 1 could be cited in the paragraphs where the family is described (Methods section, pages 3 and 4; Results section, page 5).

Discretionary revisions:

The authors discussed the potential effect of the C4329A mutation on the tRNA(Ile) and tRNA(Gln) function and showed the segregation of the mutation together with the hypertensive phenotype. Although the potential effect of this mutation is explained I would suggest if the authors have considered the possibility that this substitution could impair (or not) the cleavage and processing of the policistronic mtRNA transcripts into mature mt-tRNA species since the mutation is close to the RNAseZ cleavage site, as shown in figure 2.
**Level of interest:** An article whose findings are important to those with closely related research interests

**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.