Letter to Editors
MtDNA and Leber’s hereditary optic neuropathy

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We read with great interest the report by Li et al. concerning the association between mitochondrial DNA (mtDNA) mutations and Leber’s hereditary optic neuropathy (LHON) [1]. By clinical evaluation and genetic screening, authors identified a set of mtDNA polymorphisms in a three-generation Han Chinese family with LHON. Moreover, Li et al. regarded the mtDNA ND4 G11696A and ND5 T12338C mutations to be pathogenic mutations for LHON. However, we did not agree with several conclusions from this paper.

First, the identification of G11696A and T12338C mutations, authors used PCR-Sanger sequencing to screen the mtDNA variants in this family with LHON. However, these authors failed to notice that nuclear mitochondrial pseudogenes (NUMTs) could cause a serious problem for the mitochondrial disease studies [2], as well as sample mix-up and contamination [3]. We would suggest that a nested PCR strategy should be employed to rule out the co-amplification of nuclear pseudogenes [4].

Second, the pathogenicity of G11696A and T12338C mutations. Authors regarded the ND4 G11696A mutation to be conserved simply because this mutation was very conserved between different species, however, we noticed that the G11696A mutation was not conserved between various species, with the conservation index (CI) of 15.38% by using the Mitotool database searches [5]. In addition, the pathogenicity of human mitochondrial haplogroup F2 specific variant T12338C was still controversial [6], as this variant was also presented in the healthy subjects.

Taken together, the role of mtDNA mutation in LHON expression in this Chinese family was very problematic, a call for more carefully reassessment of the dataset remained of the approach seemed necessary.

Disclosure of conflict of interest
None.

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References


