Reviewer's report

**Title:** Association of the mtDNA m.4171C>A/MT-ND1 mutation with both optic neuropathy and bilateral brainstem lesions

**Version:** 1  
**Date:** 25 February 2014

**Reviewer:** Gregory M Enns

**Reviewer's report:**

La Morgia and colleagues have submitted a report on an interesting mitochondrial disease patient who carries the m.4171C>A mutation, and have broadened our understanding of the phenotypic spectrum associated with this specific pathogenic mutation. They also postulate that additional non-synonymous transitions affecting other ND genes may have contributed to the clinical phenotype. In addition, data on clinical course following idebenone supplementation is provided. The manuscript is interesting and well written.

**Major Compulsory Revisions**

None.

**Minor Essential Revisions**

None.

**Discretionary Revisions**

Consider adding idebenone to the key words, because there is a relative lack of data on outcomes following therapy.

It might be helpful to explain the measurement related to visual acuity, as most will not be familiar with this. Similarly, in the figure legend related to visual field a sentence or two briefly summarizing the visual field data might be considered.

**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.