Reviewer's report

Title: Transient, Recurrent, White Matter Lesions in X-linked Charcot-Marie-Tooth Disease with Novel Mutation of gap junction protein beta 1 gene in China: a case report

Version: 2 Date: 10 May 2014

Reviewer: Kleopas A Kleopa

Reviewer's report:

Major Compulsory Revisions

1. In abstract, in case presentation, and elsewhere in the manuscript the authors talk about “Demyelinating and axonal neuropathy”, however no data from nerve conduction studies are presented. Please include a table with all nerve conduction study results.

2. In abstract, in case report and in the figure legends the authors describe lesions in thalamus, which is wrong. First, thalamus is not white matter, and second, the lesion shown in the MRI figure is in the internal capsule, not in thalamus—please correct this error throughout.

3. In the background section, second paragraph, it is not correct to state in general that white matter lesions “…become the most prominent clinical manifestation” in CMT1X. They do appear in a subset of CMT1X patients, but overall peripheral neuropathy is equally prominent and present in all male patients.

4. In the background section, second paragraph, the authors write that “Cx32 maintains the permeability of neurons” This is not correct, since Cx32 is expressed by myelinating cells and not by neurons. This is also the finding of reference 2 that they correctly cite.

Minor Essential Revisions

1. In case presentation end of first paragraph, please explain “adoptive grandmother”

2. In the last page of the manuscript, second paragraph, male patient is better described as “hemizygous” rather than “homozygous” given that this is an X-linked disease.

Discretionary Revisions

1. The authors may wish to discuss the phenotype of this Cx32 novel mutation compared to the phenotype of another mutation (M93V) affecting the same amino acid.

Level of interest: An article of importance in its field
Quality of written English: Not suitable for publication unless extensively edited

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