Reviewer's report

Title: Two novel connexin32 mutations cause early onset X-linked Charcot-Marie-Tooth

Version: 1 Date: 30 March 2007

Reviewer: Angelo Schenone

Reviewer's report:

General

The authors report on two families carrying novel mutations in the gene coding for connexin32. Clinical and neurophysiological features are somehow unusual because of a relatively early onset in several family members. In general, the paper is well written and conveys interesting information to neurologists and genetists.

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Major Compulsory Revisions (that the author must respond to before a decision on publication can be reached)

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Minor Essential Revisions (such as missing labels on figures, or the wrong use of a term, which the author can be trusted to correct)

The discussion should be shortened, maybe reducing (or omitting) the notes about the frequency of abortion in female members of family 1, as a relationship with CMT is very difficult to establish. In fact, to my knowledge, there are basically no reports of increased numbers of abortions in any CMT family. Although interesting, the author should produce more evidence to conclude that abortion may be a feature of the CMTX phenotype. In fact, genetic counselling is very important in these patients and this report, lacking of a strong evidence, may be misleading.

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Discretionary Revisions (which the author can choose to ignore)

What next?: Accept after minor essential revisions

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'