Reviewer's report

Title: Spinocerebellar ataxia type 17: Haplotype analysis in pedigrees with instability of the expanded allele and absence of symptoms in a 76 year old male carrying the mutation

Version: 1 Date: 11 March 2005

Reviewer: Arnulf Koeppen

Reviewer's report:

General: Good message

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Major Compulsory Revisions (that the author must respond to before a decision on publication can be reached): The manuscript conveys three main messages: (1) case reports of an unusual family with inapparent SCA-17 but expanded CAG repeats; (2) a haplotype analysis supporting a founder effect; and (3) a comparative analysis of SCA-17 genotypes that were published before. Neither the title nor the abstract allow the reader to grasp the important conclusions. Both should be revised. The case reports on page 3 are not well rendered. If the 76-year-old father did not "accede" to further testing, how were his repeats (49) established? He must have agreed to further testing after all. The first sentence under "Conclusions" does not make sense. Also, this section is too short regarding the three-factor analysis of the paper. The headings of tables 1 and 2 should be expanded. There is no "bold" in table 1; and there is no ref. 22

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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): Reference 22 missing

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Discretionary Revisions (which the author can choose to ignore)

What next?: Unable to decide on acceptance or rejection until the authors have responded to the major compulsory revisions

Level of interest: An article of importance in its field

Quality of written English: Needs some language corrections before being published

Statistical review: No

Declaration of competing interests:

I have no competing interest