Reviewer's report

**Title:** Severe and aggressive cognitive SCA17 phenotype in a family with only marginally expanded CAG/CAA repeats in the TATA-box Binding Protein gene: A case report

**Version:** 2  **Date:** 14 May 2012

**Reviewer:** Elsdon Storey

**Reviewer's report:**

This is potentially an interesting case. I agree that the proband probably had SCA17, as a result of a minimal expansion (43 repeats). The predominantly cognitive presentation with only a minor degree of ataxia is not unusual with repeat sizes from 43-50 (see e.g. GeneTests article on SCA 17), and the reported age of onset here is average for this repeat size (ibid). The outstanding feature of this case is the reported rapidity of cognitive decline; however, as neuropsychological testing was purely cross-sectional, support for this claimed rapidity must be based entirely on history. Yet no relevant details are included. What was his educational background? What was his occupation? Is there evidence from performance appraisals that he was performing adequately in this role, say, two years before presentation? Is there other evidence of cognitive normality two years prior to presentation (e.g. no decline in intellectual hobbies, etc.)?

**Level of interest:** An article whose findings are important to those with closely related research interests

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

**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.