Reviewer's report

Title: Association between novel TARDBP mutations and Chinese patients with amyotrophic lateral sclerosis

Version: 1 Date: 5 October 2009

Reviewer: Janine Kirby

Reviewer's report:

The paper reports the first case of a TARDBP mutation in familial ALS in China. The

Minor essential revisions
1) Please provide the sex/ethnic group (Han?) of the controls.
2) Although the data suggests p.Ser292Asn is pathogenic, without functional data, you cannot state unequivocally. Thus, the abstract should be adjusted to "p.Ser292Asn is predicted to be a pathogenic mutation" and the second paragraph of the results section should have "to this pathogenic mutation" removed.
3) What do you mean by "the patient reused BiPAP"? Do you mean "refused"?
4) What did the asymptomatic father (proposed to carry the mutation) die of, and at what age?
5) Please clarify what you mean in the discussion by "None of them is homogenous"? Do you mean "homozygous"?
6) Minor corrections in the language
a) BACKGROUND - "To date, totally 30 TARDBP" would be better as "To date, a total of 30 TARDBP"
b) Clinical features - "neurological examination showed progress of disease" would be better as "neurological examination showed progression of disease"
c) Pedigree analysis - "..II5 carried Ser292Asn though his sample is unavailable to the detection" would be better as "...II5 carried the p.Ser292Asn mutation, although his sample is unavailable for confirmation"

Level of interest: An article of importance in its field

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