Reviewer’s report

Title: A patient with ALS and atypical clinical and electrodiagnostic features: a case report

Version: 2 Date: 5 June 2011

Reviewer: Shoichi Sasaki

Which of the following best describes what type of case report this is?: An unexpected association between diseases or symptoms

Has the case been reported coherently?: Yes

Is the case report authentic?: No

Is the case report ethical?: Yes

Is there any missing information that you think must be added before publication?: Yes

Is this case worth reporting?: No

Is the case report persuasive?: No

Does the case report have explanatory value?: No

Does the case report have diagnostic value?: No

Will the case report make a difference to clinical practice?: Yes

Is the anonymity of the patient protected?: Yes

Comments to authors:

This is a very interesting case in that there is a discrepancy between the clinical diagnosis and the autopsy findings: the patient has the clinical diagnosis of peripheral neuropathy, but the autopsy findings indicate the diagnosis of ALS.

To the reviewer, this case seems to have an axonal neuropathy (motor dominant) with demyelinating processes, judging from the electrophysiological findings. So, many diseases should be clinically differentiated, such as CIDP, Lewis-Summer syndrome, MMN and pseudoneuritic form of motor neuron disease.

More precise data should be needed to clarify the pathogenesis of this case.

1) This is an unusual case of ALS. More precise family history should be needed. This reviewer also recommend that the authors perform a genetic examination
including SOD1 and TDP43.

2) Was the conduction block or temporal dispersion seen by the electrophysiological study?

3) Most important, the authors should prove that the pathology of this case is authentically consistent with that of ALS, using photographs. For example, are the inclusions characteristic of ALS such as Bunina bodies, skein-like inclusions and round bodies (Lewy body-like hyaline inclusions) observed in the anterior horn cells or anywhere? How about phosphorylated TDP-43-positive inclusions? Do TDP-43-positive aggregates exist in the cytoplasm of motor neurons? Is TDP-43 immunoreactivity in the nuclei of motor neurons decreased? Vacuolar degeneration of the lateral and anterior columns is somewhat unusual for ALS pathology, and so is the corticospinal tract of the medulla. How do the authors elucidate this finding?

**Quality of written English:** Acceptable

**Declaration of competing interests:**

'I declare that I have no competing interests'