Author’s response to reviews

Title: High frequency of the TARDBP p.M337V mutation among south-eastern Chinese patients with familial amyotrophic lateral sclerosis

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Version: 2 Date: 10 Feb 2018

Author’s response to reviews:

Dear Editor and Reviewers,

We really appreciate all your comments and suggestions. Our responses are in blue below.

Reviewer reports:

Alessio Di Fonzo, Ph.D, M.D. (Reviewer 1): Authors addressed clearly all points that have been requested.

Response: Thank you.
Francisco Gondim, MD PhD FAAN (Reviewer 2): Please review language again, since there are still some linguistic problems, e.g., A physical examination could be replaced by neurological exam.

Ideally I would like to review a final version without corrections, in addition to this corrected view, so I could have a better grasp of the text, since there are too many text corrections at this point.

Response: Thank you for your careful reviewing and useful suggestions. The revised manuscript have been further edited by ‘American Journal Experts’. According to your suggestion, we uploaded the final version without corrections. The linguistic problems were corrected in the manuscript and the main linguistic changes were listed below:

1. “southeastern ” was changed as “south-eastern” in the title, abstract(line 5, line 22), introduction (line 25), methods (line 3), discussion (line 15) and conclusion section (line 2).

2. “A physical examination” was changed as “neurological exam” all through the manuscript in result section (line 18, 27, 41, 55, 63, 75, 90).

3. In abstract section (line 15), “Among the TARDBP-linked familial ALS patients, the average age of onset was 57.0±4.7 years, and a trend toward more bulbar (50.0%, 6/12) and upper limb (41.7%, 5/12) onset than lower limb onset (8.3%, 1/12) was observed.” was changed as “Among the TARDBP-linked familial ALS patients, the average age of onset was 57.0±4.7 years, and a trend towards higher rates of bulbar (50.0%, 6/12) onset and upper limb (41.7%, 5/12) onset than lower rates of limb onset (8.3%, 1/12) was observed.”.

4. In introduction section (line 5), “Typically, ALS patients initially present with symptoms in the upper limbs later in life (40-60 years old); the disease then spreads to the trunk and bulbar muscles, with patients ultimately dying from respiratory failure with a mean survival of 3-5 years [2].” was changed as “Typically, ALS patients initially present with symptoms in the upper limbs later in life (40-60 years old). The disease then spreads to the trunk and bulbar muscles, and patients ultimately die from respiratory failure, with a mean survival of 3-5 years [2]. ”.
5. In method section (line 3), “All families were recruited from the Han ethnic group from different regions of Southeastern China” was changed as “All the families were of the Han ethnic group, were recruited from different regions of south-eastern China”.

6. In method section (line 11), “Clinical profiles, including age at onset, site at onset, disease course, laboratory examination, electromyography (EMG) test, motor function, and pulmonary function, were analysed.” was changed as “Clinical profiles, including the age at onset, site at onset, disease course, laboratory data, electromyography (EMG) results, motor function, and pulmonary function, were analysed.”.

7. In result section (line 16), “However, the symptoms were not reversible and gradually worsened. Muscular weakness and atrophy spread to his upper limbs and then lower limbs.” was changed as “However, his symptoms did not resolve and gradually worsened. Muscular weakness and atrophy spread to his upper limbs and then to his lower limbs.”.