Author's response to reviews

Title: Familial Frontotemporal Dementia with Neuronal Intranuclear Inclusions is not a Polyglutamine Expansion Disease

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Author's response to reviews:

RE: Manuscript #: 1077685911107398
Title: Familial Frontotemporal Dementia with Neuronal Intranuclear Inclusions is not a Polyglutamine Expansion Disease.

Dear Editor,

The authors would like to thank the editor and the two reviewers for the insightful comments on our manuscript. The suggested changes will improve the published paper.

The following minor changes were made to the manuscript in response to the comments provided by the editor and reviewers:

1. Page 6, METHODS SECTION:

The following text was added to the methods section: 'This research was carried out in compliance with the Helsinki Declaration, and ethical approval was provided by the Clinical Research Ethics Board of the University of British Columbia (certificate C03-0449).'

2. Page 5.

In response to the reviewers query (MY), we have clarified that DNA and pathology samples from the families described in the previous publications (refs #11 and #17) are also examined in detail in the current study, and that the data presented in the current manuscript are novel. The following text was added: 'In all families studied here, the pattern of inheritance suggested an autosomal dominant trait with a high degree of penetrance as previously described [11, 17].'

3. Figure legend for Table 1:

The title of Table 1 has been modified as suggested by the reviewer to read 'CAG/CAA repeat expansions'.

4. Page 12. CONCLUSIONS:

We have clarified the conclusions as suggested (AL) and replaced the text 'most of the genes' with the following text: 'all of the predicted genes of interest'.

5. Page 11, DISCUSSION:

We have added the following text to the final sentence of the discussion as suggested by the reviewer (AL): 'although ubiquitinated intranuclear inclusions are known to occur in triplet repeat disorders encoding for amino acids other than glutamine (alanine), and in disorders caused by expansions in untranslated regions.'

Thank you for publishing our manuscript in BMC Neurology.
Yours sincerely,

Blair

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