Reviewer's report

Title: Identification of a novel functional deletion variant in the 5'-UTR of the DJ-1 gene

Version: 3 Date: 5 August 2009

Reviewer: Philipp Kahle

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The revisions satisfactorily address all concerns raised with the original manuscript. Importantly, the added patient information makes things clear now. It is a great pity that they lost the homozygous deletion bearer, who very well might have been pre-symptomatic. More, functional follow-up studies on material possibly obtained from the living patient will be very exciting.

In the final version, the authors have to add to the reference of the PD-ALS patients described by Anniesi et al. (2005) that these patients not only had the 18bp duplication, but also were double homozygous for the E163K mutation. This non-conservative exchange of an extremely well conserved acidic amino acid residue in the structurally important G-helix has been described to alter the thermal stability of the DJ-1 protein (Lakshminarasimhan et al. 2008) and alter the anti-oxidative potential of DJ-1 (Ramsey et al. 2008). Thus, it is not clear whether the Southern Italian PD-ALS patients suffered from the 18bp-duplication within the DJ-1 promoter region or the point mutation in the DJ-1 protein, or both.

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

Quality of written English: Acceptable

Statistical review: No, the manuscript does not need to be seen by a statistician.