



# THE CYPRUS INSTITUTE OF NEUROLOGY & GENETICS

## Neurogenetics Department

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30 August 2013

Dear Editor,

Please find enclosed the edited manuscript in Word format (file name: 4389-Review.doc)

**Title:** Molecular diagnosis of autosomal recessive cerebellar ataxia in the whole exome/genome sequencing era

**Authors:** Christina Votsi and Kyproula Christodoulou

**Name of Journal:** World Journal of Neurology

**ESPS Manuscript NO:** 4389

The manuscript has been improved according to the suggestions of the editor and the reviewers:

1. Format has been updated.
2. Revision has been made according to the suggestions of the reviewers as follows:
  1. Reviewer 00646254: 'This is a short and very good manuscript that provides an overview of the molecular diagnosis of autosomal recessive cerebellar ataxia in the whole exome/genome sequencing era. The originality and significance of content were very high and scientific soundness of this manuscript was also good. Thus, this manuscript can be published in world Journal of neurology after correction of some typo-errors such as technologies.'  
**Reply:** We thank the reviewer for the valuable comment. The typo-errors have been detected and corrected as highlighted in pink in the manuscript. We believe that the language of our manuscript has reached the standards set by the editorial office and does not need to be further processed for language corrections.
  2. Reviewer 02503081: 'Authors of this review article (Christina Votsi and Kyproula Christodoulou) well summarized the field on molecular diagnosis of Autosomal Recessive Cerebellar Ataxia (ARCA) emphasizing on recent discoveries observed in the whole exome/genome sequencing. However, I have minor suggestions to authors to try to include few references in Friedreich ataxia (Campuzano V, Montermini L et al Science 1996) and Also include ARCA types 1, 2 and 3 (Gros-Louis F, Dupre N, et al Nat. Genet 2007; Mollet J, Delahodde A et al Am J Hum Genet 2008; Assoum M, Salih MA et al, et al Brain 2010; Vermmer S, Hoischen A et al Am J Hum Genetic 2010). There was also a recent review paper on Autosomal Recessive Cerebellar Ataxias by Anheim M, Tranchant C, Koenig M N Engl J Med. 2012. I think it is worth mentioning this review paper'.  
**Reply:** We thank the reviewer for the valuable comments.
    1. The first reference (Campuzano et al, 1996) was already included in our references [highlighted in the reference list in blue] but might have skipped the attention of the reviewer.
    2. ARCA1, ARCA2 and rundataxin-related ataxia types and relevant references were already included in the manuscript as highlighted in yellow. We added the term ARCA3 under the

corresponding type SCAR10 on page 14 and also added the relevant references as highlighted in the manuscript and the reference list in yellow.

3. The recent review paper recommended by the reviewer has been added in the manuscript and the reference list under number 2 as highlighted in yellow.

3. References and typesetting were corrected.

Thank you again for considering our manuscript for publication in the *World Journal of Neurology*.

Sincerely yours,



Kyroula Christodoulou, PhD  
Professor of the Cyprus Institute of Neurology and Genetics  
Senior Geneticist  
Head of the Neurogenetics Department