Author's response to reviews

Title: Identification of a novel mutation in the CACNA1C gene in a Chinese family with autosomal dominant cerebellar ataxia

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Reviewer 11. The authors should perform a better clinical and neurological description of the family. For instance, what kind of saccadic pursuit abnormalities were found? Which cerebellar signs were found? How was the cognitive evaluation? The results of MOCA test should be included at the results section. Response: (1) The ocular signs of the two patients are saccadic pursuit and smooth pursuit defective. (Table 1)(2) Cerebellar signs in ULs means poor distance distinguishing and rotation dysfunction. (Figure Legends, line 18, page 11). The other cerebellar signs we have shown in Table 1.(3) We evaluated the cognition of the patients from MoCA score. (4) We have shown the MoCA score in Table 1.2. The diagnostic approach of the family should be better explained and the reason for Exome sequencing should be better described. For instance, was the genetic panel for SCA performed prior to Exome sequencing? Response: The genetic panel for SCA was performed in the proband and his son, and the common mutations and locations of SCA subtypes were excluded by fluorescence labeled capillary electrophoresis fragment analysis. And then, we chose Exome sequencing to explore the genetic defect in this family. (Pedigree and subjects, line 9-13, page 4)