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LETTER TO THE EDITOR

Reply: Cognition in SCA21 reflects developmental and adult onset cerebellar cognitive affective syndrome

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Sir,

In their Letter to the Editor, Pedro Braga-Neto *et al.* (2015) emphasized the cerebellum's major role in cognition and behaviour. The letter echoes our recent description of cerebellar impairments in patients with spinocerebellar ataxia type 21 (SCA21) caused by *TMEM240* gene mutations (Delplanque *et al.*, 2014). The affected patients' cognitive and neurobehavioural features clearly correspond to those observed in the cerebellar cognitive affective syndrome (CCAS), as first described by Schmahmann and Sherman (1998). In our study, neuropsychological assessments clearly evidenced the core features of CCAS (i.e. predominantly executive, visual-spatial, linguistic and affective changes) in three affected adults and three children from the largest SCA21 family. Most of these six patients displayed major impairments in executive function and less severe deficits in action planning, abstract reasoning and working memory abilities. Impairments in attention and in visual-spatial memory abilities were observed in the young children. Personality changes were often evidenced by impulsivity, aggressiveness, flattening of affect and rapid mood changes. Lastly, mild impairments of language were present in some children (i.e. mild anomia, limited verbal expression and some features of agrammatism). Given that most of these features appeared early in life, they are consistent with a developmental cerebellar disease. As acknowledged by Braga-Neto *et al.* (2015) it is possible that cognitive and behavioural deficits of SCA21 are related to the degeneration of focal cortical and subcortical structures in the cerebellum and in the brain. We have not yet performed neuropathological examinations of the cerebellum in the eight SCA21 families, however, we intend to use structural and functional brain imaging to (i) investigate putative dysmetria of thought; and (ii) specify the sites of the cerebellar pathology. Lastly, molecular characterization of TMEM240's function should provide us with a better pathophysiological understanding of the altered cerebellar function that characterizes the CCAS syndrome in SCA21.

References

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