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LETTER TO THE EDITOR

Spinocerebellar ataxia type 48: last but not least

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We thank Doctor Gazulla for pointing out the clinical difficulties to distinguish SCAR16 and SCA48 patients when molecular analysis does not show the presence of straightforward pathogenic variants, as in the described case [1], harboring, apparently in trans, one frameshift, with a likely impact on mRNA/protein function, and one missense, with moderate evidence of pathogenicity (meeting the PM2, PM3, PP2, PP3 ACMG criteria [2]). In our recent review [3], we retained the diagnosis of SCAR16 for the aforementioned patient (see Supplementary Table, Family P Patient 1), although we have reservations on it. Overall, this and other case presentations collected in our study [3] reinforce the impression that future functional investigations should better address how type and location of variants in *STUB1* contribute to clinical expression.

Compliance with ethical standards

Conflict of interest We have no financial interests in this manuscript.

Ethical approval None.

Human and animal rights and informed consent This reply refers to a review of the literature which did not involve human participants and did not need informed consent.

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