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Response to Commentary

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Response to Commentary *"POLG1/ANT1*-Related SANDO is a Multisystem Mitochondrial Disorder"

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We are grateful to authors of the commentary "*POLG1/ANT1*-Related SANDO is a Multisystem Mitochondrial Disorder", which highlights our previous paper "Distinctive cerebral neuropathology in an adult case of sensory ataxic neuropathy with dysarthria and ophthalmoplegia (SANDO) syndrome" [1]. Josef Finsterer's and Sinda Zarrouk-Mahjoub's comments about the well-known multi-system character in general, and cardiac complications in particular, of mitochondriopathies including SANDO are well taken, while we are not sure why the authors have concerns.

As universal cell *organellae*, dysfunctional mitochondria potentially cause a plethora of signs and symptoms. It was not our intention to dwell on this issue. Rather, we focused on the pathological substrate of the name-giving cardinal symptoms of SANDO. Thus, the main importance of our paper is the first description of the neuropathology of SANDO syndrome; this is why we chose an appropriate neuropathological journal. Like with many scientific publications today, restrictions in article volume of the respective journal, in particular in case reports, preclude description of all details that might be of any interest, including their absence. This was also the case with our report. So, a lot of questions can be always asked when patients are reported. However, many of the questions raised in the commentary are already answered in our paper (family history, clinical course, autopsy findings, absent muscle biopsy and autopsy, nerve biopsy, immunoglobulin therapy of suspected CANOMAD). The heart showed at autopsy the frequent finding of an unspecific minimal left-ventricular hypertrophy; the histology was normal. More data unfortunately have not become available. Moreover, a single case is unlikely to show all possible secondary signs and symptoms listed in the commentary. After all, SANDO syndrome was a *post mortem* diagnosis in our case.

While we fully understand, and welcome the commentary, authors intention to emphasize the multiorgan features of mitochondriopathies including SANDO, we do not feel it appropriate nor helpful to create a new term with acronym, such as the proposed mitochondrial multiorgan disorder syndrome (MIMODS).

Reference

1. Kirschenbaum D, Hedberg-Oldfors C, Oldfors A, Scherer E, Budka H. 2017. Distinctive cerebral neuropathology in an adult case of SANDO syndrome. *Neuropathol Appl Neurobiol* [In Press]. https://doi.org/10.1111/nan.12429