Author’s response to reviews

Title: Polg1 mutations and stroke like episodes: A distinct clinical entity rather by an atypical MELAS syndrome

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Dear Editors,

please find enclosed the manuscript entitled “POLG1 MUTATIONS AND STROKE LIKE EPISODES: A DISTINCT CLINICAL ENTITY RATHER BY AN ATYPICAL MELAS SYNDROME”.


A highly variable phenotypic expression is known to be associated with mutations in the POLG1 gene. Recently, a patient with a MELAS-like phenotype, carrying two heterozygous missense mutations in POLG1, has been described raising the question whether POLG1 mutations could cause MELAS or represent a distinct, even if similar, clinical entity. Here we report the case of a 79 y.o. man with a clinical phenotype highly suggestive for MELAS carrying two POLG1 mutations. Our report seem to confirm that POLG-associated phenotypes represent a distinct clinical entity, rather than an atypical MELAS syndrome.

The paper content has not been published previously elsewhere. The authors have no conflict of interests or financial support to declare. We hope that you find the manuscript suitable for publication in your journal.

Yours sincerely,
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