Replay to: Phenotypic spectrum of POLG1 mutations

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We thank Finsterer and Scorza [1] for their interest in our article.

First of all, we agree with them that any organ or tissue may be involved in mitochondrial diseases (MIDs) and that corneal epithelial dysfunction and achalasia have been described as MID manifestations [2]. In particular, these abnormalities are not very frequently reported between the POLG1 clinical manifestations.

Regarding the nerve conduction studies and progression of disease, neurophysiologic investigations have been performed in all patients. The five patients with neuropathy showed a decreased nerve conduction velocity suggestive of a mild sensory motor axonal-demyelinating peripheral neuropathy. The disease was very slowly progressive within the years. MtDNA depletion was not determined in our cases, and in Table 1 of our previous article [3], “+” and “−” indicate the presence or absence of mtDNA deletions.

Finally, for the patients with a family history negative for the disease, it was not possible to assess if the mutations were de novo or inherited because of the absence of cooperation of the other family members.

To conclude, we agree that in these diseases as well as in all the other rare neurologic disorders, a genotype-phenotype correlation, in relationship to the very large clinical heterogeneity,

is necessary by collecting many cases within a very large international collaboration. The networking within the European Academy of Neurology and the NDD-ERN will facilitate these possibilities.

Compliance with ethical standards

Conflict of interest The authors declare that they have no conflicts of interest.

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