Journal of Human Genetics (2017) 62, 867

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CORRESPONDENCE

A reply to a commentary on homozygous p.(Glu87Lys) variant in *ISCA1* is associated with a multiple mitochondrial dysfunctions syndrome

Journal of Human Genetics (2017) **62,** 867; doi:10.1038/jhg.2017.65; published online 15 June 2017

We thank Finsterer *et al.*[1] for their commentary on our article.[2] The authors comment on our paper and review other patients with multiple mitochondrial dysfunctions syndrome.

As stated in our manuscript, we investigated two families who have lost four children with this condition. Hence we could not prospectively evaluate the clinical features of these children. A review of medical records revealed clinically unremarkable respiratory, cardiac, renal, skin and endocrine status. We were unable to perform muscle biopsy and test for mitochondrial depletion as well.

We contributed our best efforts to describe the phenotype in our report (manuscript and in Table 1).[2] We speculate that retinopathy and myopathy (suggested by elevated creatine kinase) reflect multisystem manifestation of ISCA1-associated multiple mitochondrial dysfunctions syndrome, as pointed out in the commentary.[2] Isoenzymes of creatine kinase were not determined in our patients. Biochemical investigations in P2 are already provided in Table 1 and its footnote. These include renal function tests, liver function tests, and tandem mass spectrometry of blood and gas chromatography-mass spectrometry of the urine sample, which were unremarkable.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

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1 Finsterer, J. & Zarrouk-Mahjoub, S. A commentary on homozygous p.(Glu87Lys) variant in *ISCA1* is associated with a multiple mitochondrial dysfunctions syndrome. *J Hum Genet* **62**, 865–866 (2017).

2 Shukla, A., Hebbar, M., Srivastava, A., Kadavigere, R., Upadhyai, P., Kanthi, A. *et al.* Homozygous p.(Glu87Lys) variant in *ISCA1* is associated with a multiple mitochondrial dysfunctions syndrome. *J Hum Genet* **62**, 723–727 (2017).