



The Author Reply: Genetic Data Are a Prerequisite for Interpreting Clinical and Muscle Biopsy Findings in MELAS

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We would like to thank Dr. Finsterer for raising a number of important points on our paper, and agree that there were some shortcomings in our current study.

Muscle biopsy has been included in the routine analysis workup for mitochondrial disease patients if their diagnosis could not be confirmed by genetic testing. However, the usefulness of muscle biopsy, in terms of an initial diagnostic evaluation, remains unknown. In the current study, we aimed to evaluate whether muscle biopsy is useful in mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes (MELAS) patients as an initial diagnostic evaluation, and our results strongly suggested that muscle biopsy should be considered in clinically suspected MELAS patients with weakness, if not confirmed by genetic testing. Unfortunately, there has been no previous study on the usefulness of muscle biopsy as an initial diagnostic evaluation.

It is well-known that mitochondrial diseases are a heterogeneous group of disorders. Generally, the classification of a syndrome or a disease relies on accurate clinical, biochemical, and genetic information, and it may be based on either genotype or phenotype. But in cases of mitochondrial diseases, it is difficult to draw a clear line due to their heterogeneity.¹ This study was retrospective in nature, which led to several limitations. Due to its rarity, the size of our study population was small, which could have given the impression that subjects were not enough for making generalized interpretations.^{2,3} So instead,

we applied strict criteria on the patient group. Medical records of patients who were diagnosed with MELAS were reviewed based on the diagnostic criteria of Yatsuga, et al.,⁴ and patients without mtDNA mutation were excluded from the analysis.⁵

We do agree and understand Dr. Finsterer's view on the points that were raised. The study would have been more meaningful if prospective investigations could have been carried out. However, we would like to stress that a retrospective study also delivers a meaningful result despite its limitations. Again, we sincerely appreciate Dr. Finsterer's inspiring and insightful comments on this study.

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Received: March 3, 2019

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•The author has no potential conflicts of interest to disclose.

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