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Late-onset mitochondrial encephalomyopathy with lactic acid and stroke-like episodes (MELAS), defining symptomology

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With interest we read Finsterer et al.'s response [1] to our manuscript 'Case Report: 5 Year Follow-Up of Adult Late-Onset Mitochondrial Encephalomyopathy with Lactic Acid and Stroke-Like Episodes (MELAS)' [2]. We have the following responses: Regarding the lack of the MRI figure, the MRI was performed at an outside hospital. As a result an MRI figure was not available despite efforts to the contrary and only clinical interpretations were available. Upon review of the MRI report, it was determined that the patient showed a non-vascular distribution. While it remains possible the patient suffered an ischemic stroke, there is always an element of presumption in imaging-making ischemic stroke a possibility.

In response to the question of whether our patient's MELAS was indeed 'late-onset' we disagree that the treating physicians should have considered MELAS in the patient's 30s due to short stature, hypothyroidism and mild migraines, the latter of which did not require medical intervention. Whilst there may indeed be a link or associated between these symptoms and MELAS, when compared with classical MELAS cases, our patient's onset of major neurological symptoms was clearly much later. Given that short stature, hypothyroidism and hypoacusis are not enough to diagnose a patient with MELAS by defined diagnostic criteria, we find it hard to expect a physician to make a diagnosis of MELAS on these symptoms, unless they are an expert in mitochondrial disorders and even then this would be a diagnostic challenge to say the least.

For seizure treatment, the patient was given phenytoin before she was diagnosed with MELAS. Additionally, the patient requires a high dose of antiepileptic drugs because lower dosages and other therapies such as the suggested lamotrigine previously failed and resulted in sub-optimal medical management. Lastly, we agree that that all phenotype manifestations should be considered, indeed one of the purposes of our original paper was to highlight the phenotypic variability and large spectrum of disease; however, it is also important to remember clear diagnostic criteria exists.

References

[1] J. Finsterer, S. Zarrouk-Mahjoub, Onset of MELAS due to the m.3243A > G mutation is early if the large phenotypic variability is considered, Mol. Genet. Metab. Rep. 10 (2016) 23 (eCollection 2017, PMID: 27995079).
[2] K. Sunde, P.R. Blackburn, A. Cheema, J. Gass, J. Jackson, S. Macklin, P.S. Atwal, Case report: 5 year follow-up of adult late-onset mitochondrial encephalomyopathy with lactic acid and stroke-like episodes (MELAS), Mol. Genet. Metab. Rep. 9 (2016) 94–97.

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