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Status Epilepticus is a known phenotypic feature of MELAS

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LETTER TO THE EDITOR

We read with interest the article by Alenezi, *et al*. about a 28yo female with mitochondrial encephalopathy, lactic acidosis, and stroke-like episodes (MELAS) syndrome due to the variant m.3243A>G in *MT-TL1* who manifested with the classical phenotypic features of the mtDNA variant, including a status epileptics [Alenezi, A. F. *et al*., 2022]. It was concluded that status epilepticus should be considered as a manifestation of MELAS in young females with a history of stroke-like episodes (SLEs) [Alenezi, A. F. *et al*., 2022]. The study is appealing but raises concerns that should be discussed.

A limitation of the study is that heteroplasmy rates of the m.3243A>G variant were not provided. Because heteroplasmy rates can correlate with the severity of the phenotype [Yang, M. *et al*., 2021], it is crucial to determine heteroplasmy rates in clinically affected and unaffected tissues.

We disagree with the statement in the introduction that status epilepticus "has not been reported in literature reviews as a manifestation of MELAS" [Alenezi, A. F. *et al*., 2022]. On the contrary, convulsive and non-convulsive status epilepticus is a well-known phenotypic feature of the syndrome [Ribacoba, R. *et al*., 2006].

We disagree with the statement that full physical examination of the patient was non-informative [Alenezi, A. F. *et al*., 2022]. The patient had hypoacusis since age 24y, had short stature, and was underweighted [Alenezi, A. F. *et al*., 2022]. How can the physical examination be normal? We should know if underweight was due to muscle wasting in the context of myopathy.

It is reported that the index patient had a SLE prior to the current admission at age 26y and cerebral imaging [Alenezi, A. F. *et al*., 2022]. Unfortunately, there is no information about the results of the cerebral MRI at that time. We should know if the diagnosis SLE was confirmed on this MRI.

Surprisingly, the red blood cell count was elevated in the cerebrospinal fluid (CSF) [Alenezi, A. F. *et al*., 2022]. We should be told whether occult subarachnoid bleeding was considered and if a cerebral aneurysm was ruled out by computed tomography angiography (CTA).

Another limitation of the study is that the mother of the patient was not genetically tested to know if the variant m.3243A>G was inherited or occurred spontaneously. About 75% of the pathogenic mtDNA variants are transmitted via the maternal trait [Poulton, J. *et al*., 2017]. It should be also mentioned if the parents of the index patient were consanguineous or not. Knowing the genetic status of first degree relatives is crucial for assessing the outcome and for genetic counselling.

Surprisingly, creatine-kinase (CK) values were not reported [Alenezi, A. F. *et al*., 2022]. The patient had a series of epileptic seizures why hyper-CKemia can be expected. Furthermore, hyper-CKemia could result from involvement of the skeletal muscles in the disease. Mitochondrial myopathy with ragged-red fibers is a common feature of MELAS

There is no information about follow-up MRIs [Alenezi, A. F. *et al*., 2022]. To delineate whether the hyperintensities on T2/FLAIR shown in figure 1 were a postictal phenomenon or a stroke-like lesion (SLL), the morphological equivalent of a SLE, not only multimodal MRI including diffusion-weighted imaging (DWI), apparent diffusion coefficient (ADC) maps and perfusion-weighted imaging (PWI) is required but also repeated follow-ups. SLLs usually persist for weeks or even months, whereas seizure equivalents usually disappear within hours or a few days.

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Overall, the interesting study has limitations that challenge the results and their interpretation. Clarifying these weaknesses would strengthen the conclusions and could improve the study. Status epilepticus is a known phenotypic feature of MELAS. Delayed diagnosis of MELAS is the rule rather than the exception.

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