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Letter to the Editor

Suspect MELAS early to prevent unnecessary costs and burdens for the affected ☆

With interest we read the article by Trang et al. about a 34 years old female who was diagnosed with mitochondrial encephalopathy, lactic acidosis, and stroke-like episode (MELAS) syndrome upon application of the Hirano criteria. She manifested clinically with short stature, pigmentary retinopathy, hearing loss, lactic acidosis, basal ganglia calcification, dementia, recurrent stroke-like episodes at ages 27 years, 32 years, cerebellar atrophy, and 34 years respectively, and epilepsy [1]. The diagnosis was confirmed by documentation of the m.3243A>G variant [1]. It was concluded that MELAS needs to be delineated from herpes-simplex encephalitis, ischemic stroke, and posterior reversible encephalopathy syndrome [1]. The study is appealing but raises concerns.

The first limitation is that no heteroplasmy rates of clinically affected tissues were provided. Knowing heteroplasmy rates is crucial as they may determine the phenotype, the severity, and thus the outcome of the condition [2]. It would be also helpful to know if mtDNA copy number was normal or decreased in the index patient.

The second limitation is that the 3 stroke-like lesions (SLLs) were only poorly characterized on imaging. The typical features of a SLL, the morphological equivalent of a stroke-like episode on imaging, are hyperintensity on T2/fluid-attenuated inversion recovery, diffusion-weighted imaging, and perfusion-weighted imaging, hypointensity on oxygen-extraction fraction-MRI, and hypometabolism on FDG-PET [3]. SLLs most frequently originate from the cortex and spread progressively to the subcortical white matter and adjacent cortical areas. MR-spectroscopy frequently depicts an increased lactate peak, which can be confirmed by investigations of the cerebro-spinal fluid (CSF) showing elevated CSF lactate.

A third limitation is that the current medication and the treatment for the third SLL, which was correctly identified, was not provided. Knowing the treatment of the acute SLL and the chronic medication is crucial as several drugs can be mitochondrion-toxic and thus potentially deteriorate the phenotype and increase the progression of the disease. Particularly from anti-seizure drugs it is known that some can worsen the clinical presentation [4].

A fourth limitation is that no epidemiological data about mitochondrial disorders (MIDs) were discussed. It is not unusual that MELAS patients are misdiagnosed for years despite obvious manifestations suggesting the diagnosis of a MID [5]. Misdiagnosing MELAS or MIDs in general is rather the rule than the exception as most physicians are not aware of this group of metabolic disorders why they are erroneously categorized as rare or orphan disease. However, MIDs are frequent conditions which often manifest mildly at onset or mimic more common diseases. Another reason for overlooking MIDs is its frequent multisystem nature. Thus, many different specialties are involved in the management of the condition but none of them takes the lead to develop an overview and reveals the common nature of the various manifestations.

We do not agree that the CSF was normal [1]. CSF profile was abnormal as lactate was elevated.

Overall, the presented case is interesting but the report carries limitations which challenge the results and their interpretation. Concerns as outlined above need to be addressed to strengthen the conclusions.

Availability of data and material

All data reported are available from the corresponding author.

Author contribution

JF: design, literature search, discussion, first draft, critical comments.

☆ Competing interests: The authors have declared that no competing interests exist.

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