Transient symmetric T2-hyperintensities of basal ganglia and brainstem not only point to Leigh syndrome

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Full Text

Sir,

With interest we read the article by Jabeen et al. about a 37-year-old female with suspected adult-onset Leigh syndrome who profited from the administration of a mixture of thiamine, riboflavin, coenzyme-Q, L- carnitine, and L-arginine. [1] We have the following comments and concerns.

We doubt the diagnosis of adult Leigh syndrome. Arguments against Leigh syndrome are that the central nervous system lesions and the clinical manifestations completely resolved, that no mtDNA mutation was detected, that the family history was negative for a mitochondrial disorder (MID), and that ragged-red fibers were absent on muscle biopsy. Immunohistological findings on muscle biopsy could be secondary, as well as elevated serum lactate and the magnetic resonance spectroscopy findings. Bilateral T2-hyperintense lesions of the thalamus, [2] the periaqueductal grey, [3] the hypothalamic region [3] have also been reported in patients with thiamine deficiency. The patient did not undergo lactate stress testing or investigations for mutations in nuclear DNA-encoded genes.

Which were the results of the biochemical investigations of the muscle homogenate? Was there isolated Complex I, Complex II, Complex III, Complex IV deficiency or multiple complex deficiencies as has been previously reported in Leigh syndrome? [4],[5] Which were the muscle biopsy findings on electron microscopy? Was there cristae destruction, swelling of mitochondria, intramitochondrial glycogen deposition, or paracrystalline inclusions?

Which was the cause of abdominal pain and vomiting since 3 months? Did the patient suffer from gastrointestinal pseudoobstruction as has been reported in patients with Mitochondrial neurogastrointestinal encephalomyopathy syndrome. [6] Was aseptic pancreatitis, repeatedly reported as a manifestation of MIDs, the cause of her complaints?

Which was the cause of breathlessness and shock when the patient was readmitted shortly after dismissal? Which type of shock was diagnosed, cardiac or septic? Could an unwitnessed seizure or self-limiting status epilepticus (e.g., aborted sudden unexpected death in epilepsy) explain metabolic acidosis or lactate elevation? Was an electroencephalogram recorded during this episode? Was creatine kinase, troponine-T, or proBNP elevated? Which were the results of the electrocardiogram and echocardiography at that time? Were there any indications for acute heart failure due to a Takotsubo syndrome, which also could explain the acute deterioration? Was the cause of respiratory insufficiency cardiac, infectious, or muscular? For how long did the patient require artificial ventilation?

Overall, the presented data strongly argue against an MID. MIDs are usually progressive in nature and only marginally respond to vitamin/cofactor cocktails. The only MID, which potentially responds to such compounds is primary coenzyme-Q deficiency, [7] which often responds favorably to coenzyme-Q, and should be excluded in this interesting patient as well.

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Conflicts of interest

There are no conflicts of interest.

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