Response to the letter to the editor

Sir,
I thank Mahjoub *et al*. for showing interest in our article about a patient with suspected Leigh's disease.

We arrived at the diagnosis of mitochondrial disease and in particular Leigh's disease in a 37-year-old female due to the following reasons.

In her first presentation, the patient presented with unexplained vomiting followed by acute brainstem syndrome, which was diagnosed as Wernicke's encephalopathy in view of the horizontal gaze-evoked nystagmus and ataxia with imaging evidence of dorsal brainstem lesions. In the first admission, investigations to evaluate for the cause of recurrent vomiting were done including endoscopy and colonoscopy, which were normal. Retrospectively, the reason for unexplained gastrointestinal (GI) symptoms[2] is probably mitochondrial disease as the patient is completely asymptomatic after using mitochondrial regimen (thiamine and coenzyme Q).

The patient stopped thiamine after discharge and she relapsed back with severe symptoms. The shock was not due to sepsis as the total white blood cell count and serum procalcitonin including blood and urine cultures were normal. A normal central venous pressure with normal two-dimensional echo, lack of elevation of creatinine kinase, and troponin levels also rules out the possibility of takotsubo cardiomyopathy or any cardiac cause for the ventilatory failure or shock.[1] The most plausible cause for respiratory failure was brainstem involvement as she had associated bulbar palsy. The patient was fully conscious even in the presence of severe lactic acidosis making the possibility of underlying status epilepticus less likely. Moreover, the patient improved without any antiepileptic drug.

We agree with Mahjoub *et al*. that the investigations to establish the molecular diagnosis of mitochondrial disease were incomplete due to unavailability of nuclear DNA analysis for mitochondrial genes and electron microscopy. Regarding the course of the disease in adult-onset Leigh's disease as they pointed out, it ranges from sudden death shortly after onset to recovery and long-term survival.[2,3] Our patient, at the time of this writing, is doing well and continuing to take coenzyme Q and thiamine supplements. Although the molecular diagnosis could not be established, the clinical profile of the patient (unexplained GI symptoms, unexplained elevation of serum and cerebrospinal fluid lactate, imaging picture, supportive features on muscle biopsy, the dramatic response to thiamine and other mitochondrial cocktail strongly favors a mitochondrial disease, most likely Leigh's disease.[4]

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Conflicts of interest
There are no conflicts of interest.

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