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Letter to the Editor

Before Attributing Respiratory Failure in Mitochondrial Myopathy to Respirator Muscle Weakness, Alternative Causes must be Ruled Out

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Letter to the Editor

We read with interest the article by Hoang *et al*. about a 7-year-old boy with progressive mitochondrial myopathy due to the m.3251A>G variant in the MT-TL1 gene. The boy developed acute muscle weakness and respiratory failure, initially attributed to myositis, which necessitated intubation and mechanical ventilation [1]. Despite the administration of glucocorticoids and intravenous immunoglobulin (IVIG), the muscle disease progressed, prompting a muscle biopsy that confirmed the suspected mitochondrial myopathy [1]. The study is interesting, but some points warrant discussion.

First, it is unclear why dermatomyositis was initially suspected [1]. Were the inflammatory markers in the blood (CRP, erythrocyte sedimentation rate, leukocyte count) elevated? Myositis-specific and myositis-associated antibodies were negative, and the patient showed no dermatological abnormalities typical of dermatomyositis [1]. On admission, the patient's serum lactate level was elevated, suggestive of mitochondrial disorder (MID). Why was he not initially tested for other features of MID? Furthermore, mitochondrial myopathy can be associated with muscular respiratory insufficiency and contrast enhancement on muscle MRI due to secondary inflammation [2].

Second, a cause of respiratory insufficiency in the central nervous system (CNS) was not sufficiently ruled out [1]. Since the patient had lactic acidosis, elevated lactate levels in the brain and cerebrospinal fluid (CSF) cannot be excluded. Lactic acidosis in the CNS can also impair respiratory function [3], which is why lactic acidosis in the CSF should have been ruled out by direct lactate measurement in the CSF or by magnetic resonance spectroscopy (MRS).

Third, MIDs are generally multisystem diseases [4]. Therefore, it should be clarified whether the patient was systematically examined for involvement of organs other than skeletal muscle. In addition to CNS involvement, as in the present patient, patients with mitochondrial myopathy can also have ocular, otological, endocrine, cardiac, gastrointestinal, renal, or peripheral nerve involvement.

The fourth point is that an MRI without evidence of a stroke-like lesion (SLL) does not necessarily rule out the possibility that the patient has never previously experienced a stroke-like episode (SLE). Since SLLs can disappear completely without leaving any visible structural abnormality on cerebral imaging [5], we should be informed whether the patient had a history of SLE prior to admission for respiratory failure.

The fifth point is that no family history was provided [1]. Since MIDs are inherited maternally in 75% of cases due to mtDNA mutations [6], it should be reported whether the mother or other first-degree relatives were also affected by MID. It should also be clarified whether the patient's mother also carried the pathogenic MT-TL1 variant.

The sixth point is that the use of steroids without a confirmed diagnosis of immune myositis is surprising. Since steroids can have severe adverse effects, including death, particularly in patients with MID [7], they should be used with caution.

Overall, patients with a MID should generally be evaluated for multi-organ involvement. In cases of lactic acidosis, the initial diagnostic workup should include an assessment for possible MID. Before respiratory insufficiency in mitochondrial myopathy is attributed to respiratory muscle weakness, alternative causes must be ruled out.

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