Mitochondrial myopathy, dysmorphism, exercise-induced vomiting and tachycardia the mutation m.4831G > A

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Letter to the Editor,

With interest we read the article by Zanolini et al. about a 21 year-old male with isolated myopathy due to the ND2 mutation m.4831G > A [1]. We have the following comments and concerns.

We do not agree that the patient had myopathy as the sole manifestation of the mitochondrial disorder (MID). The patient started with vomiting during exercise since age 17 years, a frequent manifestation of MIDs particularly in patients with MELAS and possibly associated with lactic acidosis [2]. Additionally, he had bone abnormalities, manifesting as high arched palate and malocclusion of teeth [1]. Furthermore, exercise-induced supraventricular tachycardia suggests cardiac involvement. The patient thus has to be classified as mitochondrial multiorgan disorder syndrome (MIMODS) [3].

Particularly at onset of the disease, the multisystem nature of a MID may not be clinically evident. In such a case active and prospective search for MIMODS has to be carried out. What were the results of pulmonary, endocrine, gastro-intestinal, renal, hematological, and dermal investigations?

We also do not agree with the statement that the neurological exam was normal [1]. The patient is described with diffuse muscle wasting and reduced tendon reflexes [1]. Additionally, abnormal endurance tests were reported [1]. Thus, the neurological exam was indicative of a peripheral nervous system problem, and not normal.

Since the patient had developed exercise-induced vomiting, we should be informed if he had a history of migraine, cycling vomiting syndrome, or a gastrointestinal problem. Was the history positive for stroke-like episodes, frequently associated with vomiting among other manifestations [4]. Though cerebral lactate was normal on "MRI", most likely MR-spectroscopy, we should be informed if the patient had undergone cerebro-spinal fluid investigations to rule out elevated cerebral lactate. Lactate determination on MRS may be normal despite the presence of cerebral lactic acidosis.

Overall, this interesting case should be prospectively investigated for MIMODS and the cause of exercise-induced vomiting should be clarified.

References


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