Letter to the Editor

Antiepileptics and NO-precursors may be beneficial for stroke-like episodes

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ABSTRACT

Pathogenesis and management of stroke-like episodes in mitochondrial disorders is under debate and no consensus has been reached thus far on how this phenomenon should be managed. Frequently applied are nitric oxide (NO) precursors but a well-designed study confirming the effectiveness of such an approach is lacking.

A shortcoming of the report is that no heteroplasmy rates were provided. Since heteroplasmy rates may strongly influence the severity of the phenotype, it is essential to know the mutation load in affected tissues.

The patient was obviously underweighted with 32 kg and a body height of 149 cm [1]. Was the low body weight attributable to a gastrointestinal abnormality, to a specific diet the patient was taking regularly, to a hormonal problem, or to the drugs she was regularly taking? Did the patient suffer from a depressive disorder? Since mitochondrial disorders are more frequently associated with neoplasias than healthy subjects [5], we should be informed if a malignoma was definitively excluded in this particular patient. Gastrointestinal involvement in MELAS that could explain the low body weight is well appreciated and includes vomiting [6], pseudoobstruction, poor appetite, gastroesophageal sphincter dysfunction, constipation, dysphagia, gastroparesis, diarrhoea, pancreatitis, or stenosis of the duodeno-jejunal junction [7]. Patients with a MID are frequently recommended to adhere to a ketogenic diet to avoid glucose intake and stimulation of the glycolytic pathway. We should be informed if the presented patient was under such a diet or any other dietary regimen that could explain the low body weight. Was reduced body weight simply due to growth hormone deficiency, a rare endocrine manifestation in MIDs [8]. Since the ketogenic diet may exhibit beneficial effects in MELAS in general [9] and in mitochondrial epilepsy in particular [10], we should be informed if the patient was put on such a regimen.

A shortcoming of the report is that no heteroplasmy rates were provided. Since heteroplasmy rates may strongly influence the severity of the phenotype, it is essential to know the mutation load in affected and unaffected tissues.

Overall, this interesting case requires a broader discussion of the MRI findings, an explanation why antiepileptics were given and why no ketogenic diet was recommended, and provision of heteroplasmy rates in hair follicles, buccal mucosa cells, skin fibroblasts, muscle cells, lymphocytes, and urinary epithelial cells.
Conflicts of interest

There are no conflicts of interest.

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References


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