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Letter to the Editor

Ogilvie Syndrome can Mimic Intestinal Pseudo-Obstruction in m.3243A>G Carriers

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Letter to the Editor

We read with interest the article by Liu *et al*. about a 24-year-old patient with MELAS syndrome (mitochondrial encephalopathy with lactic acidosis and stroke-like episodes) due to the m.3243A>G variant in the MT-TL1 gene with a heteroplasmy rate of 61% and TRPM3-related neurocognitive impairment due to the heterozygous c.2878G>T variant in the TRPM3 gene [1]. The patient presented clinically with confusion, generalized tonic-clonic seizures, intestinal pseudo-obstruction (IPO), lactic acidosis, and bilateral lesions of the basal ganglia, cerebellum, and thalamus [1]. She benefited significantly from jejunal decompression [1]. The study is interesting, but some questions remain.

First, it was not clearly distinguished between the clinical manifestations attributable to the m.3243A>G variant and those attributable to the TRPN3 variant [1]. Assigning clinical manifestations to one variant or the other is crucial, as treatment, course, and outcome may differ. TRPN3-related neurodevelopmental disorder manifests clinically with intellectual disability, developmental delay, epilepsy, and hypotonia [2]. Mutations in the TRPM3 gene are classified as gain-of-function mutations that lead to overactive, calcium-permeable cation channels [2].

Secondly, MELAS is diagnosed according to either the Hirano or the Japanese criteria [3, 4]. According to the Japanese criteria, MELAS is diagnosed when there are signs of encephalopathy associated with dementia or epilepsy, stroke-like episodes (SLE) in early childhood, and biochemical evidence of mitochondrial dysfunction, such as acidosis and the presence of ragged-red fibers in muscle biopsy [4]. According to the Hirano criteria, MELAS is diagnosed when SLE occur before the age of 40 and there are seizures or dementia, lactic acidosis or ragged-red fibers, normal early childhood development, recurrent headaches, or recurrent vomiting [3]. For these reasons, MELAS syndrome should not be diagnosed in the index patient. Since the index patient had no history of a stroke-like episode (SLE), which is the pathognomonic feature of MELAS, the clinical diagnosis remains unclear. Given that the patient presented with bilateral symmetrical lesions of the basal ganglia and thalamus, a diagnosis of Leigh syndrome should be considered rather than MELAS.

Third, IPO is not a rare phenotypic feature in MELAS. It is attributed to the involvement of smooth muscle cells with respiratory chain dysfunction, leading to reduced peristalsis or even complete cessation of intestinal contraction activity [5]. IPO has been described in several patients with the m.3243A>G variant [6, 7, 8, 9].

Fourth, no immunohistological or biochemical analyses were performed on the jejunal sample obtained during decompression surgery. Since the reduced contractility of the smooth muscle cells is most likely due to smooth muscle cell involvement, it would have been interesting to know whether the functions of the respiratory chain complexes are also reduced in the jejunal smooth muscle cells.

Fifth, it cannot be ruled out that the IPO was caused by treatment with tranquilizers as antiepileptic drugs. This phenomenon is known as Ogilvie syndrome and can also be caused by drugs with strong anticholinergic properties or those that affect the autonomic nervous system. In addition to benzodiazepines, clozapine and olanzapine can also trigger Ogilvie syndrome [10].

In summary, patients with MELAS should meet the Hirano or Japanese criteria. Symmetrical lesions of the basal ganglia, thalamus, and cerebellum are more indicative of Leigh syndrome than MELAS. IPO is not a rare manifestation of the m.3243A>G variant but has been repeatedly described. Before attributing IPO in an m.3243A>G carrier to the underlying gene defect, Ogilvie syndrome must be ruled out.

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