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Letter to the Editor

Before Diagnosing Superior Mesenteric Artery (Wilkies) Syndrome in MELAS, other causes of Gastroparesis must be ruled out

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We read with interest the article by Horna *et al*. about a 22-year-old male with mitochondrial encephalopathy, lactic acidosis, and stroke-like episode (MELAS) syndrome clinically presenting with stroke-like episodes, epilepsy, pancreatitis, perimyocarditis, gastroparesis, vomiting, constipation, and superior mesenteric artery syndrome (SMAS) [1]. SMAS was managed conservatively, which had a beneficial effect, so that the patient regained weight through feeding via home parenteral nutrition (HPN) [1]. The study is impressive, but some points require further discussion.

The first point is that the underlying mutation responsible for MELAS has not been reported [1]. We should know whether MELAS in the index patient was due to an mtDNA defect or a mutation in a nuclear mitochondrial gene. Since >90% of MELAS cases are due to mtDNA mutations and 80% of patients carry the m.3243A>G variant, it is very likely that MELAS in the index patient was also due to the m.3243A>G variant. When an mtDNA defect is suspected, it is important to report the heteroplasmy rate of the causative variant in order to assess the genotype-phenotype correlation.

The second point is that no family history was provided. To assess whether MELAS syndrome was inherited or occurred sporadically, it is important to know whether other first-degree relatives, especially the mother, were also affected and whether any of the affected relatives carried the same mutation as the index patient.

The third point is that gastroparesis could also be due to autonomic denervation or smooth muscle myopathy. How was parasympathetic under-innervation ruled out? Did the patient undergo biopsy of the gastric smooth muscle to determine whether smooth muscle myopathy was present or not? What were the results of intestinal biopsy? Did the patient undergo a ^{13}C-acetate breath test to determine half-emptying time (HET) and peak time of the ^{13}C-%-dose-excess curve (T max), or was the oro-cecal transit time (OCTT) evaluated by analysing $^{13}CO_2$ exhalation curves after ingestion of ^{13}C labelled test meals?

A fourth point is that no duodenoscopy or duodenography has been reported to document duodenal obstruction.

A fifth point is that current medications were not provided. Since medications can cause or promote gastrointestinal problems, it is important to know what the patient was taking regularly. Apparently, he was taking antiepileptic medication on a long-term basis. What other medications did he take regularly?

A sixth point is that the patient was diagnosed with "perimyocarditis" [1], which is an unusual phenotypic feature of MELAS. MELAS has been reported to manifest as hypertrophic cardiomyopathy [2] or dilated cardiomyopathy [3], but not myocarditis. Was the myocarditis due to infection or due to immune mechanisms? Was myocarditis diagnosed by cardiac MRI and documentation of late gadolinium enhancement (LGE), or was myocarditis diagnosed by endo-myocardial biopsy?

In summary, the interesting study has limitations that put the results and their interpretation into perspective. Before gastroparesis in MELAS can be attributed to SMAS, alternative etiologies must be thoroughly ruled out.

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